Alberta Congenital Anomalies Surveillance System
Eighth Report: 1980 - 2007

Alberta

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Government of Alberta

Suggested citation: Alberta Health and Wellness (2009). *Alberta Congenital Anomalies Surveillance System: Eighth Report 1980 - 2007.* Edmonton, AB: Alberta Health and Wellness.

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ISBN 978-0-7785-8247-2 Print
ISBN 978-0-7785-8248-9 Online

ALBERTA CONGENITAL ANOMALIES SURVEILLANCE SYSTEM

EIGHTH REPORT

1980 – 2007

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Acknowledgements

The Alberta Congenital Anomalies Surveillance System (ACASS) receives funding from Alberta Health and Wellness (AHW) for the on-going collection of data on all congenital anomalies in Alberta. ACASS is located at the Alberta Children's Hospital in Calgary and has received in kind support from the Calgary Health Region. The success of ACASS also depends upon the interest and activities of many people including hospital Health Records personnel, unit clerks, nurses, clinic co-ordinators and physicians. Many physicians are contacted by letter in order to obtain additional clarifying information and their prompt replies are appreciated.

Following agencies and individuals are acknowledged for their contribution to the production of this report:

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ACRONYMS USED IN THE REPORT

ACASS	Alberta Congenital Anomalies Surveillance System
CCASN	Canadian Congenital Anomalies Surveillance Network
ICBDSR	International Clearinghouse for Birth Defects Surveillance and Research
NBDPN	National Birth Defects Prevention Network

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1. EXECUTIVE SUMMARY

1. This is the eighth in a series of reports detailing the prevalence of congenital anomalies in Alberta particularly dealing with years 2003-2007 inclusive. Aggregate data are also included from 1980.

2. The International Classification of Diseases – 10th Edition (ICD-10) classification system has been adopted by Alberta and the report uses the Royal College of Pediatrics and Child Health adaptation of ICD-10. The anomalies outlined in the National Birth Defects Prevention Network's Guidelines for Conducting Birth Defects Surveillance (**http://www.nbdpn.org/index.html**) are reported in this report. However, all items from the ICD-10 "Q" codes as well as other sections such as disorders of metabolism are monitored by ACASS. Data on such disorders will be provided to interested parties upon request.

3. Congenital anomaly rates have remained relatively stable over the years with fluctuations occurring on a year to year basis. There are, however, notable exceptions such as neural tube defects, Down syndrome, and gastroschisis.

4. The implementation of folic acid fortification of flour in 1998 resulted in 46% decrease in neural tube defect rates in Canada. It also brought all provincial rates to the same level where previously there had been a markedly higher rate in Eastern Canada compared to Western Canada with BC and Alberta having had the lowest rates (New Eng J Med 2007 357:143-153). There was an increase in spina bifida in 2006 which is currently unexplained but the rate has now returned to its former levels.

5. Gastroschisis continues to show an increase and is particularly prevalent in young mothers, which is consistent with worldwide observations from other jurisdictions. Omphalocele on the other hand shows no such increases, nor is there an association with low maternal age. In fact there is a higher frequency of omphalocele found in higher maternal ages, that is 40 years of age and older.

6. The increase in Down syndrome is likely attributable to the increased number of women giving birth aged 35 years or older. A number of other congenital anomalies are associated with advanced maternal age including ventricular septal defects, nervous systems defects, renal dysgenesis and congenital hip dislocation.

7. The percentage of births to women 35 years of age and over continues to increase with approximately 16% of women in this age category giving birth in the period 2003-2007.

8. The total number of births in Alberta has increased from 36,860 in the year 2000 to 48,683 in 2007.

9. In Alberta about 19% of infants with congenital anomalies were low birth weight in 2007 which is consistent with our previous report.

10. The birth prevalence of congenital anomalies has varied by health region, which is partly due to regional variations in case ascertainment and reporting. With the merging of health regions it will become more difficult to investigate regional variations.

11. Alberta data were used to investigate the possible increase of two congenital anomalies which could not have been easily investigated in other provinces. The studies done in Alberta (anophthalmia/microphthalmia and anal atresia) showed that the increases were not statistically significant (Can J Ophthalmol 2005 40:38-44; J Ped Surg 2007 42:1417-1421).

12. ACASS continues to be a member of the Canadian Congenital Anomalies Surveillance Network (CCASN), a Public Health Agency of Canada initiative, with members of ACASS playing a significant role in Network committees. The Network has been formed to support the development and maintenance of high quality population-based surveillance systems for congenital anomalies.

13. ACASS continues its affiliation with the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) and has participated in group studies in a number of congenital anomalies including craniofacial defects, very rare defects, gastroschisis, holoprosencephaly and Down syndrome ascertainment. Publications on these are listed in the Appendix.

2. INTRODUCTION

This Report provides updated data on congenital anomalies ascertained in Alberta from the years 1980-2007 inclusive. For the current release, the anomalies outlined in the National Birth Defects Prevention Network's (NBDPN) Guidelines for Conducting Birth Defects Surveillance (2004) are reported, however, data on other anomalies can be provided upon request.

2.1 History

The history of the Alberta Congenital Anomalies Surveillance System (ACASS) has been well described in previous reports. Since 1996, funding has been provided by Alberta Health and Wellness, Surveillance and Assessment Branch. ACASS continues to work closely with Alberta Vital Statistics and relies on them for the provision of notifications of births, deaths and stillbirths.

2.2 Purpose of a Surveillance System

Public health surveillance in general has been defined by the Centers for Disease Control and Prevention (CDC) in Atlanta, Georgia as the ongoing, systematic collection, analysis and interpretation of data (e.g. regarding agent/hazard, risk factor, exposure, health event) essential to the planning, implementation and evaluation of public health practice, closely integrated with the timely dissemination of these data to those responsible for prevention and control.

The purposes and objectives of surveillance for congenital anomalies (CAs) are to:

1) provide reliable and valid baseline data of congenital anomalies in Alberta;
2) investigate any significant temporal or geographic changes in the frequency of congenital anomalies with a view to identifying environmental, and therefore, possibly preventable causes;
3) measure trends;
4) assess the effectiveness of prevention (e.g. folic acid fortification or antenatal screening);
5) assist with health related program planning and development through the provision of data;
6) participate in research into the etiology and natural history of birth defects;
7) assist research through provision of congenital anomalies data; and
8) provide advice to health care professionals about congenital anomalies, especially with respect to teaching and launching public health campaigns (e.g. folic acid campaign by Community Health in Calgary).

As well as the above, patterns or associations of malformations to determine whether they belong to an existing or new syndrome complex can be explored.

A principle feature of a surveillance system is timeliness; however data collection and analysis should not be accomplished at the expense of an accurate diagnosis. Since data are collected by the 1st birthday, plus the possibility of delay in reporting, the data of a given calendar year may not be complete until at least the December 31 of the subsequent year.

2.3 Prevention of Congenital Anomalies (Birth Defects) – Can We Do Better?

There is increasing concern about environmental contaminants and many questions have been raised about relationships between environmental pollutants and health problems such as respiratory illness, cancer or birth defects. It is therefore of the utmost importance to keep accurate databases on the occurrence and geographical distribution of illness such as cancer or birth defects which in the latter case is the task of ACASS. There is no current indication of any geographical or environmental clustering of birth defects in Alberta but that does not mean that surveillance should cease. On the contrary, surveillance for trends or clustering of birth defects is a safeguard for the population.

As far as temporal effects and trends are concerned these are also reasonably stable with three exceptions; two of which involve increases and one a decrease. The decrease is in the prevalence of neural tube defects especially anencephaly compared to 10 years ago and is likely the result of folic acid fortification and/or the use of folic acid tablets preconceptionally and in the first trimester of pregnancy. The increases have occurred in gastroschisis and Down syndrome and for very different reasons. Younger maternal age is associated with gastroschisis and older maternal age with Down syndrome. The increase in gastroschisis is a worldwide phenomenon and Alberta is no exception. Gastroschisis occurs mainly in infants of very young mothers (15 to 19 yrs), to a lesser degree the increase also occurs among the 20 to 24 yr age group with relative stability in prevalence among subsequent maternal ages. While the exact cause is unknown there is a correlation with a number of risk factors such as the absence of prenatal care, use of vasoconstrictive recreational drugs such as ecstasy, amphetamines and cocaine, smoking cigarettes or marijuana, poor nutrition and genitourinary infections. Down syndrome in contrast has a much higher frequency in infants of older mothers (35 yrs and over), a fact known for many years. The change in our population is the increasing tendency for women to defer childbearing until later in their biological life span. About 16% of all mothers are age 35 years or over compared to 4% over the past two to three decades.

Birth defects prevention strategies have been known and implemented with varying degrees of success over the years, examples of which include Rubella immunization, improved glycemic control for diabetic mothers and avoidance of alcohol, tobacco and drugs that are known to be teratogenic (e.g. thalidomide, antiepileptic, and isotretinoins). Indeed other drugs, even if not proven to be teratogenic, should be avoided in pregnancy if possible. If needed, they should be used only after careful re-assessment by a physician. More recently, the use of folic acid in the prevention of neural tube defects has contributed to their reduction. Two additional measures could further reduce the frequency of birth defects; reduction of maternal obesity and multivitamin and folic acid supplementation.

Maternal Obesity

There is substantial evidence that birth defects are more frequent among obese mothers (BMI ≥ 30) and even for overweight mothers (BMI 25.0 – 29.9) than among normal weight (BMI 18.5 – 24.9) women. The increase in birth defects involves many different systems including neural tube defects, heart defects, both septal and conotruncal, orofacial clefts, limb reduction defects, obstructive urinary tract defects and anorectal atresia.

Multivitamins and Folic Acid

The second simple public health measure to reduce the prevalence of birth defects would involve campaigns to increase the use of multivitamins and folic acid prior to conception and throughout the first trimester of pregnancy. Folic acid is already in flour in Canada, the United States of America and Chile. Whether folic acid alone reduces selected birth defects is controversial but it does seem clear that multivitamins with or without folic acid supplements do seem to have a definite preventive effect on a variety of congenital anomalies including those of the cardiovascular system, obstructive defects of the urinary tract and orofacial clefting. Not only would some of the above measures reduce the frequency of birth defects but would also be likely to reduce the frequency of preterm births and low birth weight infants.

Summary

Public health campaigns are needed to educate the population on these simple measures to reduce the prevalence of birth defects. To evaluate the results there is a clear need to maintain accurate and up to date perinatal and congenital anomaly databases. It has taken approximately 50 years for the majority of the population to stop smoking hence it is clear that such campaigns are not short term projects. We should strive to educate the public so that most pregnancies are planned and have access to optimal periconceptional/prenatal care. Reduction of birth defects will result in improved birth outcomes and reduce the burdens of illness upon the individual, the parents and family but will also result in cost savings across many sectors including health, education and employment.

Selected Bibliography

Botto LD, Olney RS, Erickson JD. 2004. Vitamin Supplements and the Risk for Congenital Anomalies Other Than Neural Tube Defects. Am J Med Genet Part C:125C, 12-21.

Czeizel AE, Dobó M, Vargha P. 2004. Hungarian Cohort-Controlled Trial of Periconceptional Multivitamin Supplementation Shows a Reduction in Certain Congenital Abnormalities. Birth Defects Res A 70:853-861.

Czeizel AE. 2009. Periconceptional folic acid and multivitamin supplementation for the prevention of neural tube defects and other congenital abnormalities. Birth Defects Res A: Clinical & Molecular Teratology. Online pub 22 Jan 2009.

Goh YI, Bollano E, Einarson TR et al. 2006. Prenatal Multivitamin Supplementation and Rates of Congenital Anomalies: A Meta-Analysis. J Obst Gynecol Can 28:680-689.

Johnson CY, Little J. 2008. Folate intake, markers of folate status and oral clefts: is the evidence converging? Int J Epidemiol 37: 1041-1058.

Mastroiacovo P. 2008. Risk factors for gastroschisis: Genitourinary infection in early pregnancy can be added to the existing list. BMJ 336:1386-1387.

Oddy WH, De Klerk NH, Miller M et al. 2009. Association of maternal pre-pregnancy weight with birth defects: evidence from a case-control study in Western Australia. Austral & New Zealand J Obst & Gynaecol 49:11-15.

Stothard KJ, Tennant PWG, Bell R et al. 2009. Maternal Overweight and Obesity and the Risk of Congenital Anomalies: a systematic review and meta-analysis. JAMA 301:636-650.

Watkins ML, Rasmussen SA, Honein MA et al. 2003. Maternal Obesity and Risk for Birth Defects. Pediatrics 111:1152-1158.

Wilcox AJ, Lie RT, Solvoll K et al. 2007. Folic acid supplements and risk of facial clefts: national population based case-control study. BMJ 334:464-469.

3. METHODOLOGY

3.1 Case Definitions

A **congenital anomaly** is an abnormality that is present at birth, even if not diagnosed until months or years later. Most congenital anomalies are present long before the time of birth, some in the embryonic period (up to the end of the 7th week of gestation) and others in the fetal period (8th week to term). The term "anomaly" covers all the major classes of abnormalities of development, of which there are four major categories as follows:

Malformation – a morphologic defect of an organ, part of an organ or a larger region of the body resulting from an intrinsically abnormal developmental process (e.g. spina bifida, cleft lip and palate).

Deformation – an abnormal form, shape or position of a part of the body caused by mechanical forces (e.g. extrinsic force such as intrauterine constraint causing some forms of clubfoot).

Disruption – a morphologic defect of an organ, part of an organ or a larger region of the body resulting from the extrinsic breakdown of, or an interference with, an originally normal developmental process (e.g. an infection such as rubella or a teratogen such as thalidomide).

Dysplasia – the abnormal organization of cells into tissues and its morphologic result (e.g. Marfan Syndrome, osteogenesis imperfecta).

Other definitions related to pregnancy outcomes for the purposes of this report are as follows:

Live birth – a complete expulsion or extraction from the mother, *irrespective* of the duration of the pregnancy, of a foetus in which, after expulsion or extraction, there is breathing, beating of the heart, pulsation of the umbilical cord or definite movement of voluntary muscle (Alberta Vital Statistics Annual review, 2000).

Stillbirth – a complete expulsion or extraction from the mother, after at least 20 weeks pregnancy (≥20 weeks), *or* after attaining a weight of 500 grams or more (≥500 grams) of a fetus in which, after the expulsion or extraction, there is no breathing, beating of the heart, pulsation of the umbilical cord or unmistakable movement of voluntary muscle (Alberta Vital Statistics Annual review, 2000).

Gestation – completed weeks of pregnancy at delivery.

Preterm birth (aka premature) – a birth before 37 weeks of gestation (<37 weeks).

Termination of Pregnancy (ToP) – for our purposes, any pregnancy loss before 20 Weeks gestation (<20 weeks), most of which are therapeutic terminations for congenital anomalies but could include spontaneous abortions or intrauterine fetal deaths with fetal anomalies.

3.2 Case Ascertainment

An infant can be ascertained at any time up to the first birthday. Multiple ascertainment of the same infant can occur and is encouraged, as this frequently improves the quality and reliability of the data.

As several malformations may occur in the same infant, it is advantageous to allow each to be reported so that groups of associated malformations may be studied. This, however,

leads to difficulties since the final tabulations may be reported as total malformations (anomaly rates) or as the total number of malformed infants (case rates).

ACASS obtains information about infants with congenital anomalies from a variety of independent sources. Acquisition of additional reporting agencies is always a priority since the use of multiple sources of information improves not only the ease but also completeness of ascertainment as well as for verification of the diagnostic data. **Appendix A.1** shows the process of data collection at ACASS.

ACASS screens many important Alberta Health and Wellness and Alberta Vital Statistics documents for the presence of a congenital anomaly including:

- Notice of a Live Birth or a Stillbirth and Newborn Record often referred to as the Physician's Notice of Birth (NOB)
- Medical Certificate of Stillbirth
- Medical Certificate of Death

Also, ACASS screens a notification called the Congenital Anomalies Reporting Form (CARF, **Appendix A.2**) that is completed by all acute care hospitals in the province on live births, stillbirths, admissions or hospital deaths of infants under one year of age as well as pregnancy losses involving one or more congenital anomalies. This form serves as the single most important source of case ascertainment.

Since many children with congenital anomalies are not admitted to hospital, it is very important to obtain out-patient information such as from the Calgary and Edmonton Departments of Medical Genetics.

Ascertainment at a continued high level requires each hospital record department and each health care provider to co-operate with the system by notifying us as promptly as possible. We are fortunate in having such co-operative agencies and personnel.

3.3 Quality Control Measures

When a copy of a reporting document reaches the ACASS office in Calgary, it is reviewed for content by the Research Assistant and Manager. If the information is unclear, the Manager, on behalf of the Medical Consultant, writes to the physician responsible for the case seeking clarification. A stamped, addressed envelope is included with the letter and the physician is asked to respond at the bottom of the letter thus making the mechanics of replying easy. The response from physicians has been very satisfactory (greater than 90%) and usually this is sufficient to make a decision whether to accept or reject an anomaly or case. Any questionable diagnosis that is not confirmed is not entered into the database. Some cases also not included contain diagnoses that do not belong in a congenital anomaly system or are part of a normal developmental process such as a patent ductus arteriosus or undescended testes in a premature infant. Any reports requiring a medical decision are reviewed with the Medical Consultant. Policy decisions with respect to the acceptance or rejection of a case and its coding are referred to the ACASS Advisory Committee. This body is comprised of a paediatric cardiologist, neonatologist/epidemiologist, paediatric pathologist, medical geneticist (medical consultant) with occasional input from a paediatric neurologist, paediatric nephrologist and a paediatric orthopaedic surgeon.

3.4 Anomaly Coding

Coding is done at the Calgary office using the Royal College of Paediatrics and Child Health (RCPCH) adaptation of the International Classification of Diseases, tenth edition (ICD-10). Difficult cases are referred to the Medical Consultant (Medical Geneticist). In the past, we were able to code only 6 anomalies per case but since 1997 we have been coding all eligible anomalies reported to us.

3.5 Data Linkage

Data from ACASS are linked to data from the Alberta Vital Statistics Birth Registry by the birth registration number, with over 99% success. Some maternal risk factor data, such as maternal smoking, drinking and use of street drugs during pregnancy are thus available for babies with congenital anomalies. This linkage enables in-depth data analysis and interpretation.

3.6 Confidentiality and Release of Data

Notifications of Congenital Anomalies are sent to the Surveillance and Assessment Branch, Alberta Health and Wellness and from there to the ACASS office in Calgary where the database is maintained. The notifications are handled by the Manager, Research Assistant, Secretary, Clerk and Medical Consultant. The data are treated in a completely confidential manner and the notifications are kept in locked files in a locked room. The database is secured by limited access and is password protected. Should further clarification about a case or anomaly become necessary, we communicate with the attending physician or the physician responsible for ongoing care. Direct contact is never made with the family. When data are requested from us, they are released in aggregate form with no personal identifiers.

3.7 Epidemiological and Statistical Measures

Unless otherwise stated, the birth defect rates presented in this report are calculated using the following formulae:

$$\text{ANOMALY (DEFECT) RATE} = \frac{\text{Number of a particular congenital anomaly among live births and stillbirths}}{\text{Total number of live births and stillbirths}} \times 1000$$

$$\text{CASE RATE} = \frac{\text{Number of individual infants (live- or stillborn) with} \geq \text{1 congenital anomaly}}{\text{Total number of live births and stillbirths}} \times 1000$$

Confidence intervals (95 per cent) are also included because the rate obtained is actually only a point estimate of the unknown, true population rate. The confidence interval provides information about the precision of the estimate. Thus, the confidence intervals are an estimated range of values within which there is a 95% probability that the true population rate will fall.

Linear trend analysis was performed and presented as appropriate.

3.8 Limitations of Data and Analysis

One of the major limitations of the surveillance system is that on its own, the information provided to us does not allow studies to determine etiology. If increasing trends indicate there is a potentially serious problem, then separate investigative studies need to be done. However, it is possible to conduct linkage studies with other data sources to explore potential causes of specific birth defects.

The ACASS data are collected passively from Vital Statistics, hospitals, and other agencies but are augmented by active ascertainment from physicians and labs, etc. The completeness and accuracy of data are largely dependent on reporting. Regional variations in anomaly and case rates may be explained in part by ascertainment and reporting differences. As such, caution has to be exercised in the interpretation and comparison of regional data.

Some anomaly groups have a small number of cases thus are combined to allow stable rate estimations and meaningful comparisons. The specific methods used for each section are included in the last portion of the section.

4. PATTERNS OF SELECTED CONGENITAL ANOMALIES IN ALBERTA

4.1 Birth Prevalence – Time Trends

The following table and graphs of selected sentinel anomalies indicate the trends in congenital anomaly rates in Alberta from 1980 through 2007. Sentinel anomalies are those which the International Clearinghouse of Birth Defects Surveillance and Research (ICBDSR), of which we are a member, watches worldwide with the rationale that they are quite easily identified hence accurately reported.

Table 4.1 Chi Squared Linear Trend Analysis and p-values for Selected Anomalies 1980-2007 Inclusive (Live Births and Stillbirths)

Anomaly	Trend Direction	Chi Squared Analysis (χ^2LT)	p-value
Neural Tube Defects	Decreasing	36.09	0.0000
Anencephaly	Decreasing	37.15	0.0000
Spina Bifida	Decreasing	11.46	0.0007
Hydrocephalus	No significant change	0.02	0.8875
Cleft Lip +/- Cleft Palate	No significant change	1.74	0.1871
Cleft Palate	No significant change	1.06	0.3032
Oesophageal Atresia/Stenosis	Decreasing	4.08	0.0434
Anorectal & Large Intestine Atresia/Stenosis	No significant change	1.57	0.2102
Hypospadias and Epispadias *	No significant change	0.10	0.7518
Limb Reductions	Decreasing	18.11	0.0000
Gastroschisis	Increasing	57.80	0.0000
Omphalocele	No significant change	0.27	0.6033
Down Syndrome	Increasing	63.47	0.0000
Renal Agenesis	Increasing	4.69	0.0303
Hypoplastic Left Heart Syndrome	Increasing	5.42	0.0199

*Hypospadias and Epispadias calculated for male births only

Table 4.2 presents a comparison of birth prevalence rates between ACASS and a selection of other countries or regions as reported in the International Clearinghouse for Birth Defects Surveillance and Research Annual Report, 2007.

Table 4.2 Selected Anomaly Rates for Alberta and Other Jurisdictions Reporting to the ICBDSR, 2001-2005* Rates per 1000 Total Births, ToPs included.

Anomaly	Alberta	Western Australia	Atlanta	Utah	Wales	Texas (2001-2004)
Neural Tube Defects	0.70	1.43	0.72	0.73	1.60	0.68
Anencephaly	0.23	0.58	0.21	0.23	0.65	0.25
Spina Bifida	0.31	0.73	0.36	0.41	0.76	0.35
Hydrocephalus	0.57	0.76	0.65	0.22	0.82	0.60
Cleft Lip +/- Cleft Palate	1.25	1.30	0.87	1.32	1.08	1.06
Cleft Palate	0.78	1.17	0.52	0.76	0.93	0.52
Oesophageal Atresia/Stenosis	0.21	0.42	0.21	0.26	0.18	0.20
Anorectal Atresia/Stenosis	0.64	0.68	0.30	0.36	0.39	0.49
Hypospadias	2.08	3.19	0.73	0.44	2.00	1.59
Limb Reductions	1.15	0.65	0.42	0.66	0.85	0.52
Gastroschisis	0.39	0.36	0.28	0.53	0.62	0.42
Omphalocele	0.24	0.48	0.17	0.24	0.44	0.21
Down Syndrome	2.13	2.62	1.73	1.56	2.06	1.26
Renal Agenesis	0.60	0.49	0.09	0.37	0.37	0.19
Hypoplastic Left Heart Syndrome	0.26	0.14	0.23	0.36	0.34	0.20

* www.icbdsr.org/filebank/documents/ar2005/Report2007.pdf

4.1.1 Neural Tube Defects

Neural tube defect rates have declined in Alberta since 1980 especially anencephaly and spina bifida. Encephalocele rates have remained fairly constant. Terminations of affected pregnancies and early fetal losses might account for part of the decline but it is unlikely that these factors alone explain the total reduction of NTDs in Alberta. We have been able to follow terminations of pregnancy (ToPs) and early fetal losses since 1997. However, if we extrapolate our ToP data back through the years, there seems to be a true decline in the rates of NTDs since the 1998 introduction of folic acid fortification of flour and cereal/grain products (150 µg/100gm, **Figure 4.1.1, 4.1.2**). We participated in the seven-province study of the impact of folic acid fortification and the prevalence of neural tube defects. The results showed a decrease of 46% in the overall rates and brought all provincial rates to the same level whereas previously there was a markedly high rate in eastern Canada compared to the west (NEJM 2007, 357:143-153). After many years of decline there was however a striking increase in spina bifida in 2006 followed by a dramatic fall in 2007 back to the new lower level established since 1999. The spike in 2006 is unexplained and did not include anencephaly nor can it be accounted for by an increase in syndromic forms of spina bifida. Our data were also used in an International Clearinghouse study showing that lipomyelomeningocele prevalence showed a slight but no statistically significant decrease in this anomaly following folic acid fortification (Birth Defects Res A 2008, 82:106-109).

Figure 4.1.1 Neural Tube Defects in Total Births (Live + Still), Alberta, 1980-2007

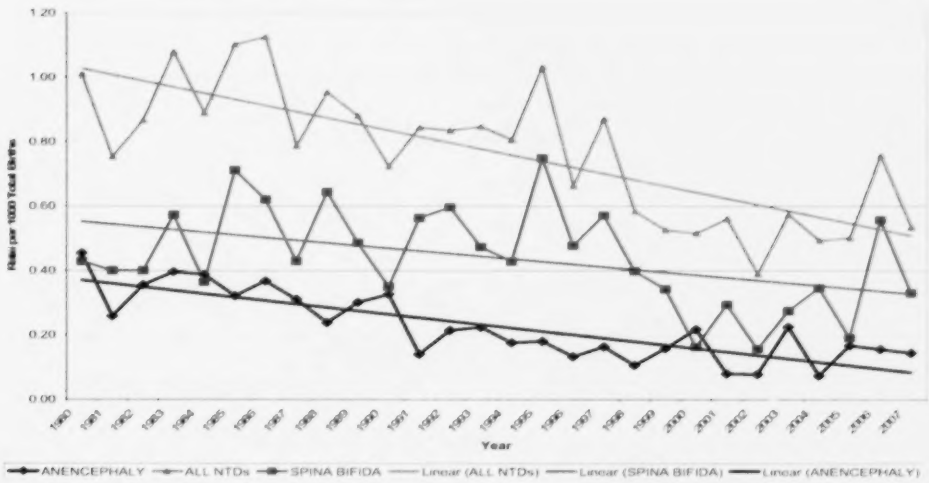
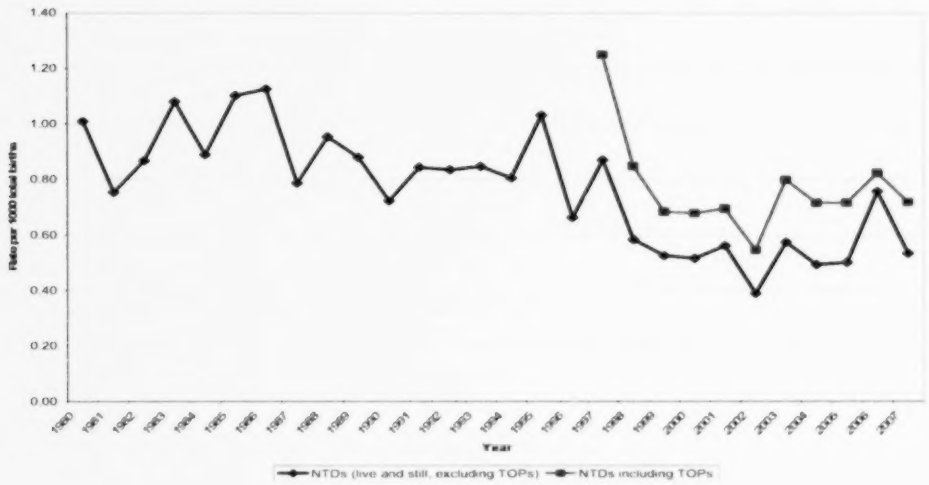


Figure 4.1.2 Neural Tube Defects including ToPs/Fetal Losses, Alberta, 1980-2007



4.1.2 Cleft Lip and Palate

The birth prevalence of cleft lip with or without cleft palate (CL ± CP) remains stable (**Figure 4.1.3**). There are so few ToPs in either group that their addition is not enough to alter the trends in either case (CL ± CP or CP alone). These results suggest that the introduction of folic acid fortification has had no affect on the rates of either of the clefting groups although there are some studies suggesting that a reduction has occurred following fortification. The reduction might be due to multivitamins with or without folic acid, but whether folic acid alone is effective is controversial (Int J Epidemiol 2008, 37:1041-1048; BMJ 2007, 334: 464-469).

Cleft lip with or without cleft palate has never shown the secular changes that neural tube defects showed. In fact CL ± CP and CP rates have been stable for many years – in British Columbia from 1952-86 (CMAJ 1989, 140:1167-1170) and in Alberta from 1980-2002 (CCASN Current Contents Winter 2005 (http://www.phac-aspc.gc.ca/ccasn-rcsac/ct2005/or-cl-alberta-eng.php)).

Figure 4.1.3 Cleft Lip +/- Cleft Palate (CL+/-CP) and Cleft Palate (CP) Alone in Total Births (Live + Still), Alberta, 1980-2007

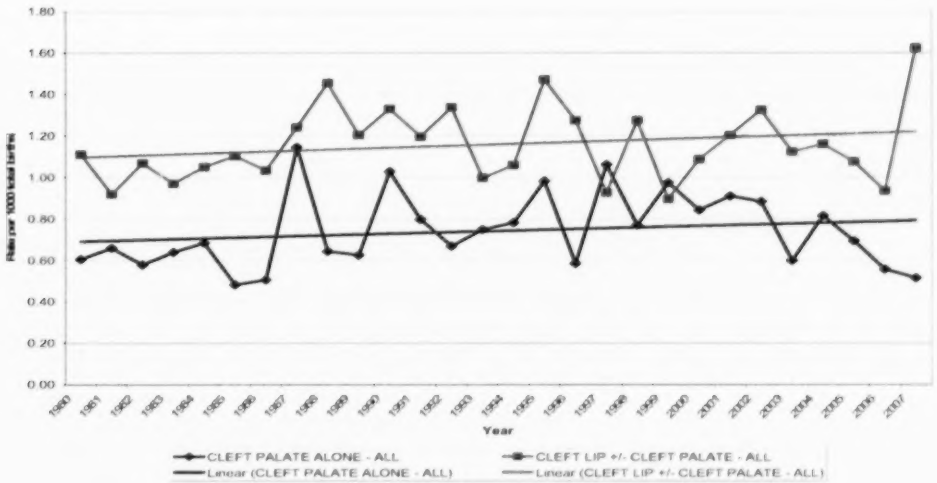


Figure 4.1.4 Cleft Lip +/- Cleft Palate in Total Births (Live + Still), Alberta 1980-2007 Isolated and Total Excluding Syndromes 1990-2007

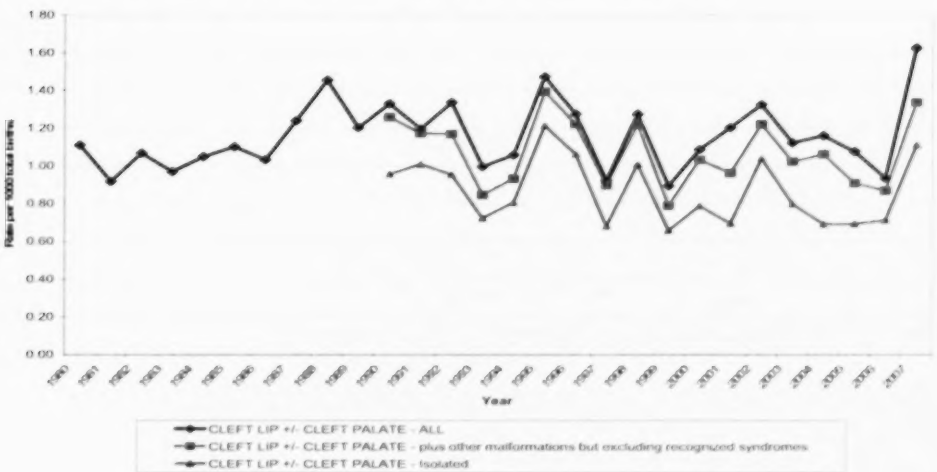
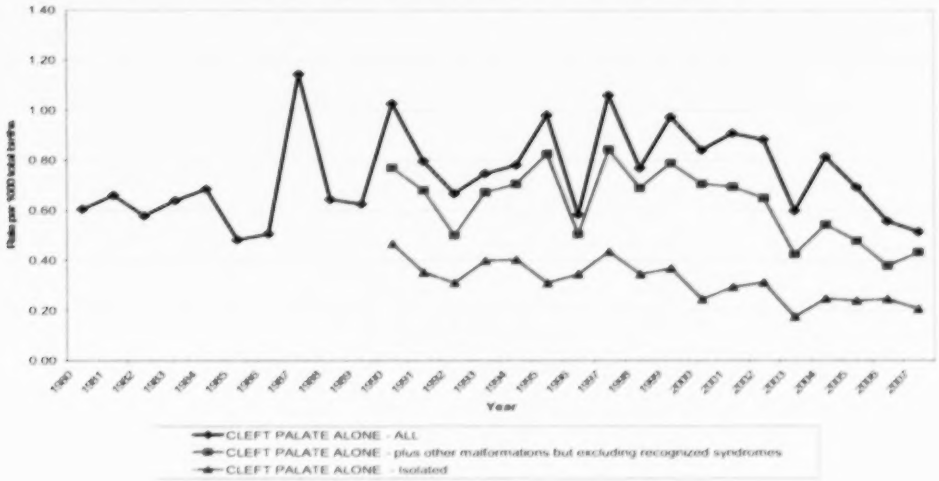


Figure 4.1.5 Cleft Palate Alone in Total Births (Live + Still), Alberta 1980-2007 Isolated and Total Excluding Syndromes 1990-2007



4.1.3 Abdominal Wall Defects

These defects include mainly gastroschisis and omphalocele. Increasing gastroschisis rates are a world-wide phenomenon occurring particularly in younger mothers, that is, under 20 years of age, and to a lesser extent in the 20 to 24 year age group. In contrast, omphalocele annual prevalence rates are relatively constant however they show a reverse maternal age effect in that there is a marked increase in the 35 to 39 year maternal age group and more so in the 40 and over age group. Our data were also included in a publication from the International Clearinghouse (BMJ 2006, 332:423-424) (**Figure 4.1.6-4.1.9**).

Figure 4.1.6 Abdominal Wall Defects – Gastroschisis and Omphalocele in Total Births (Live + Still), Alberta, 1980-2007

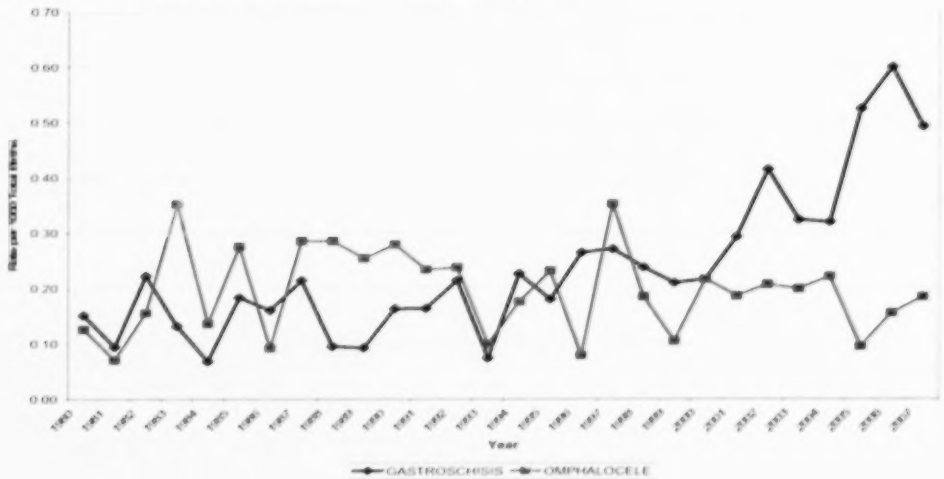


Figure 4.1.7 Gastroschisis by Maternal Age Groups in 5 Year Increments (except 2005-2007) Alberta, 1980-2007

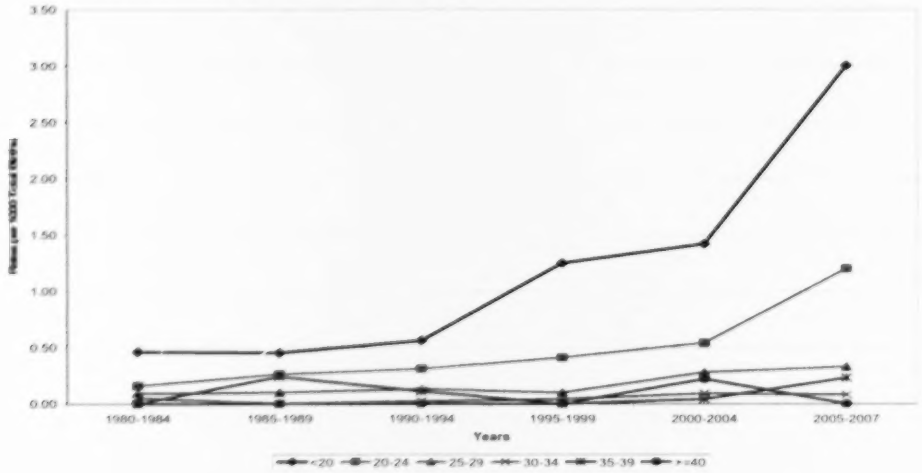


Figure 4.1.8 Gastroschisis With and Without ToPs/Fetal Losses, Alberta, 1980-2007

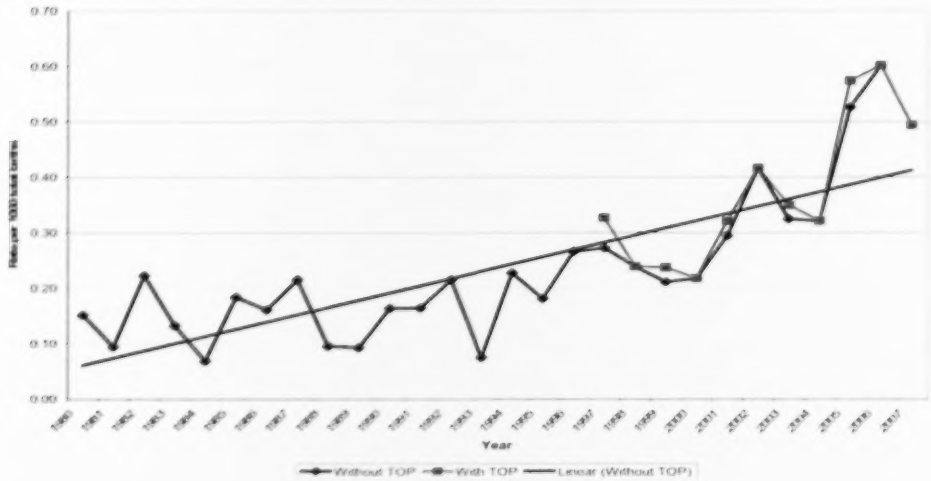
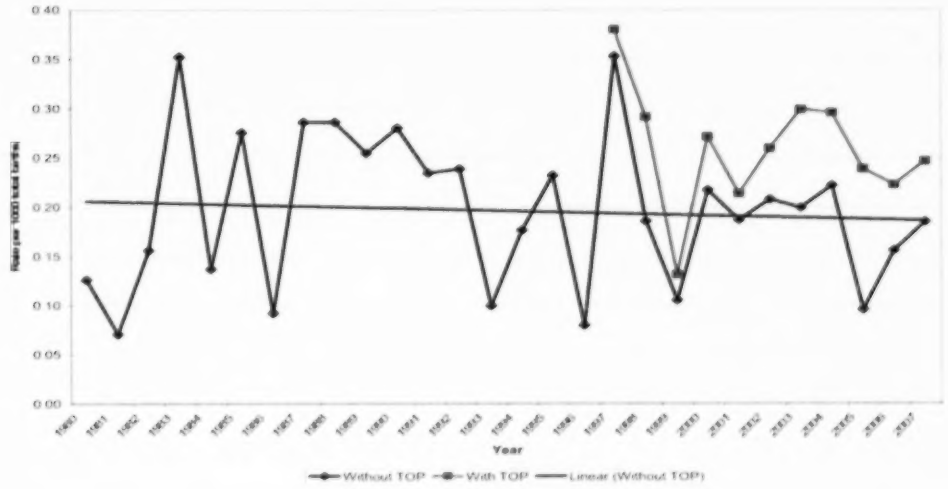


Figure 4.1.9 Omphalocele With and Without ToPs/Fetal Losses, Alberta, 1980-2007



4.1.4 Chromosome Anomalies

From 1980-2007 there were 2455 chromosomal anomalies reported to ACASS. Of these, 1779 (74%) were either Trisomy 13 (Patau Syndrome), Trisomy 18 (Edwards Syndrome) or Trisomy 21 (Down Syndrome). Down Syndrome was by far the most commonly ascertained chromosome anomaly – 78% of the above mentioned group of trisomies and 58% of the total number of chromosome anomalies reported. Sex chromosome anomalies accounted for approximately 7% of the total.

As previously reported, Down Syndrome rates are increasing (**Figure 4.1.10**) but are strongly correlated with increasing maternal age. In 1980, 4% of mothers were 35 years of age or over whereas in 2007, 16% of mothers were in the same age category.

Table 4.3 Down Syndrome Rates per 1000 Total Births (Live + still) By Maternal Age, 2003-2007 With Rates Including ToPs in Brackets

Maternal Age	Year 2003	2004	2005	2006	2007
<20	1.87 (1.87)	0.47 (0.47)	1.38 (1.84)	0.00 (0.00)	0.80 (0.80)
20-24	0.87 (0.87)	1.01 (1.01)	0.50 (0.62)	0.69 (0.81)	0.55 (0.76)
25-29	0.97 (0.97)	0.71 (0.78)	1.37 (1.52)	0.85 (0.99)	0.84 (1.03)
30-34	1.30 (1.74)	1.19 (1.53)	1.65 (2.06)	0.92 (1.30)	1.51 (1.94)
35-39	4.42 (5.63)	5.80 (6.80)	4.91 (6.61)	3.48 (5.22)	3.63 (6.15)
≥40	11.81 (20.67)	6.99 (10.98)	14.18 (20.79)	9.13 (17.35)	13.20 (21.45)
All ages	1.79 (2.29)	1.68 (2.02)	2.05 (2.65)	1.33 (1.94)	1.66 (2.38)

Figure 4.1.10 Chromosome Anomalies: Trisomy 13, Trisomy 18, Trisomy 21, in Total Births (Live + Still), Alberta, 1980-2007

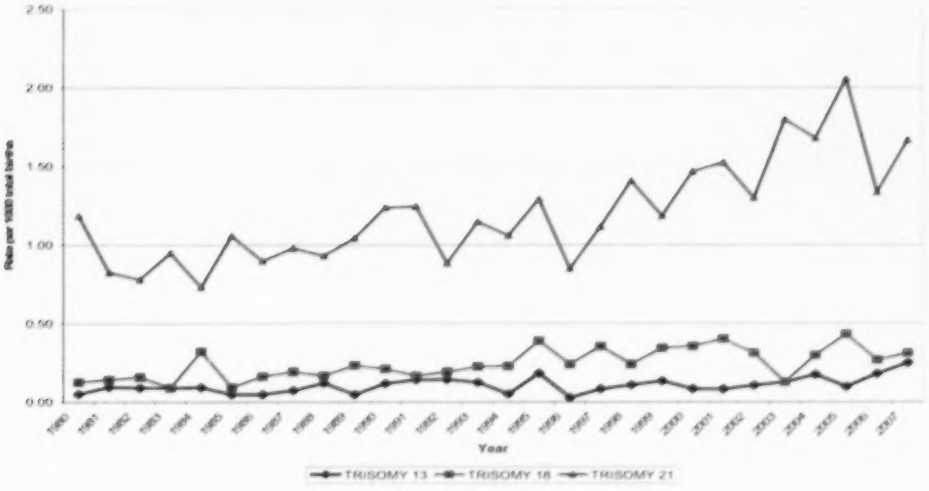
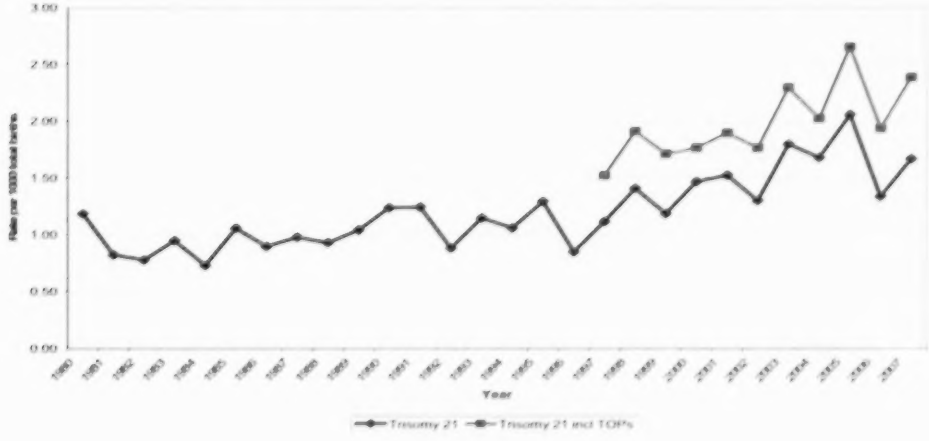


Figure 4.1.11 Down Syndrome With and Without ToPs/Fetal Losses, Alberta, 1980-2007



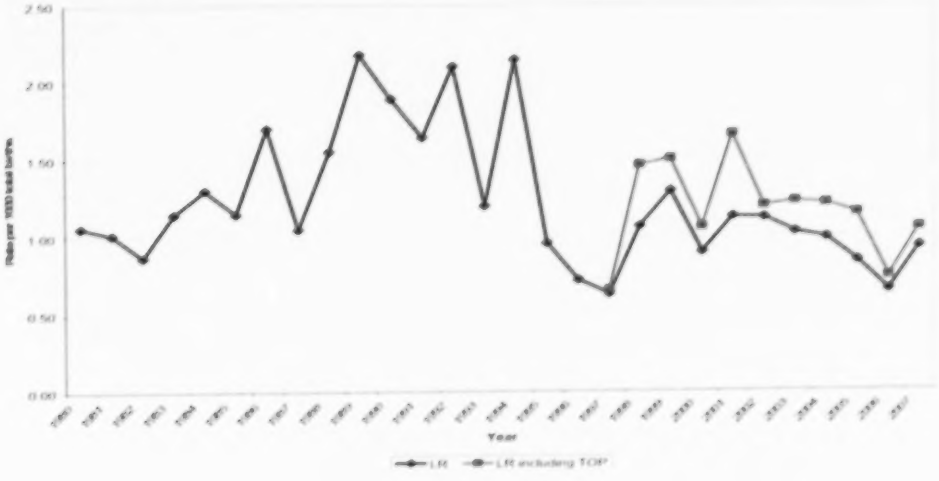
Terminations of pregnancy (ToPs) do not affect rates of Down Syndrome markedly until we examine births to women over the age of 30 (**Figure 4.1.11, Table 4.3**). As mentioned earlier in the report, ACASS has collected data on ToPs since 1997. Table 4.3 demonstrates the rates of Down Syndrome by maternal age for the years 2003 to 2007. The rates including ToPs are in brackets.
ACASS data were included in an ICBDSR study of ascertainment of Down Syndrome which showed that we compared favourably to many other surveillance systems (manuscript in press).

Infants with Down Syndrome often have associated anomalies. ACASS does not code minor anomalies associated with Down syndrome such as single palmar crease, upslanting palpebral fissures, and increased space between the first and second toes. However, other major malformations, if mentioned on the ascertainment documents, are entered routinely into the database.
Assessment of ascertainment efficacy for Down Syndrome was undertaken by members of the ICBDSR which showed that ACASS has a very good rate of ascertainment (ICBDSR – submitted for publication).

4.1.5 Limb Reductions

After some years of increasing trends prior to 2000, prevalence rates have declined (**Figure 4.1.12**). ACASS is also participating in an international study, co-ordinated through the ICBDSR, on the epidemiology of very rare defects among which are some of the more uncommon limb reduction defects such as true phocomelia (absence of all limb bones proximal to the hand or foot - the hand or foot attaching directly to the trunk) and amelia (complete absence of one or more limbs).

Figure 4.1.12 Limb Reduction Defects With and Without ToPs/Fetal Losses, Alberta, 1980-2007



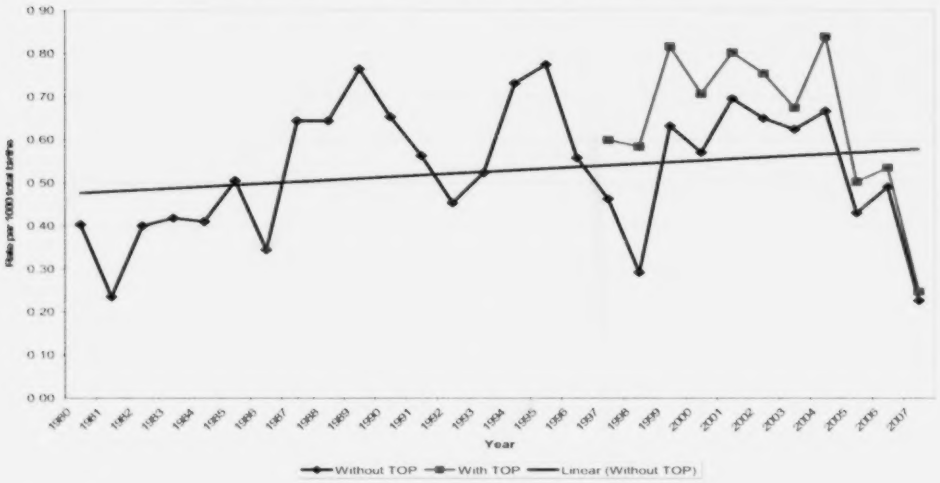
4.1.6 Anorectal Atresia/Stenosis

There has been no significant change in the prevalence rates (**Figure 4.1.13**). The rates which appeared to increase in 1998 in both Alberta and Canadian National data prompted a detailed survey of Alberta data. The results which were subsequently published (J Peds Surg 2007, 42:1417-1421) showed no overall trend for both all cases isolated and multiple combined. The results indicated that a substantial number of cases belonged in the multiple congenital anomaly VATER/VACTERL.

Our prevalence rates are very comparable to two other large population studies, one from British Columbia and the other from the EUROCAT registries with frequencies in the 1/2200

– 1/2500 range (AJMG 1986, (suppl 2): 151-161; ibid 2001, 103:207-215 and 2002, 110:122-130).

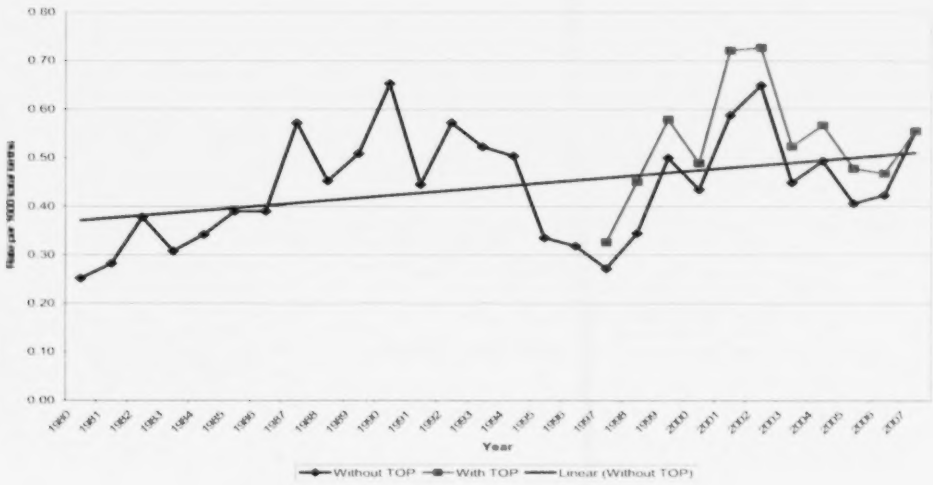
Figure 4.1.13 Anorectal and Large Intestine Atresia/Stenosis With and Without ToPs/Fetal Losses, Alberta, 1980-2007



4.1.7 Renal Agenesis/Hypoplasia

These rates continue to show an increasing trend which could be due to better ascertainment as a result of improving diagnostic capabilities (**Figure 4.1.14**).

Figure 4.1.14 Renal Agenesis/Hypoplasia With and Without ToPs/Fetal Losses, Alberta, 1980-2007

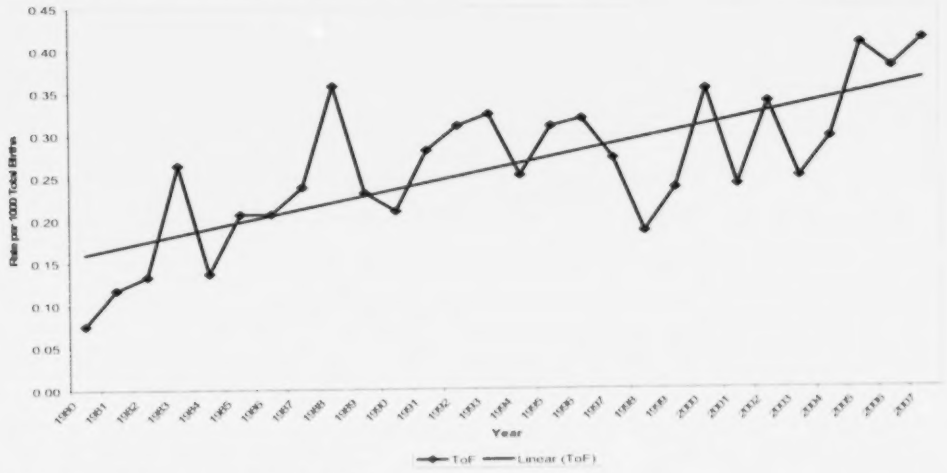


4.1.8 Congenital Heart Disease

Despite the likelihood of improved ascertainment, two defects that appear to be showing a steady increase over time are Tetralogy of Fallot (**Figure 4.1.15**) and Hypoplastic Left Heart Syndrome (**Figure 4.1.16**). There is also a slight upward trend for septal defects both atrial and ventricular. With the assistance of the two major pediatric cardiology facilities in the province (Drs. Joyce Harder and John Dyck) ACASS personnel are undertaking a Quality Assurance Study of congenital heart disease for the years 1995-2001. A secondary objective of the study is to see whether folic acid fortification has made any difference to congenital heart defect rates. Recent publications suggest that the combination of multivitamins and folic acid taken periconceptionally may reduce the prevalence of septal and conotruncal defects. There could be a number of reasons why these prevalence rates are increasing such as;

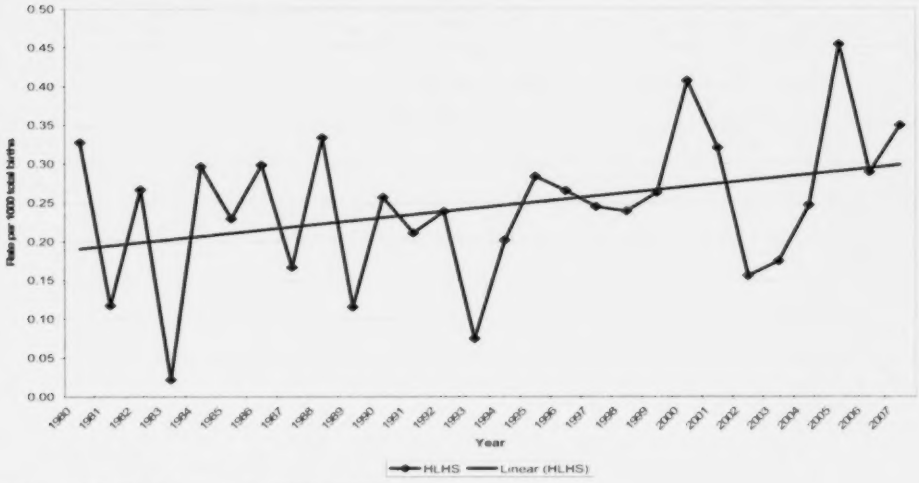
a) improvement in ascertainment and diagnosis, and
b) lack of appropriate timing for ingesting multivitamins and folic acid such as delaying ingestion until pregnancy is confirmed.

Figure 4.1.15 Tetralogy of Fallot in Total Births (Live + Still), Alberta, 1980-2007



Note: 1 ToP for years 1997-2007

Figure 4.1.16 Hypoplastic Left Heart Syndrome in Total Births (Live + Still), Alberta, 1980-2007



Note: 5 ToPs for years 1997-2007

4.1.8 Summary

ACASS reviews anomalies that have been entered into the database on a regular basis. As is evident from the review of anorectal atresia and neural tube defects, detailed studies of some individual anomalies or anomaly groups aid in the assessment and maintenance of the data quality. With intensive review, some cases might be re-assigned, re-coded or discarded altogether from the database. This continuing review might explain some discrepancies in the data from earlier reports.

5 SURVEILLANCE AND RESEARCH PROJECTS

5.1 Surveillance and Research Projects/Collaborations and Consultations/Papers

1. De Wals P, Tairou F, Van Allen MI, Uh SH, Lowry RB, Sibbald B, Evans J, Van den Hof MC, Zimmer P, Crowley M, Fernandez B, Lee NS, Niyonsenga T. Impact of folic acid food fortification on the prevalence of neural tube defects in Canada. New Eng J Med. 357:143-153, 2007.
2. Lowry RB, Sibbald B and Bedard T. Stability of prevalence rates of anorectal malformations in the Alberta Congenital Anomalies Surveillance System 1990-2004. J Pediatr Surg 42:1417-1421, 2007.
3. Paquette D, Lowry RB and Sauvé R. Two to three percent of infants are born with a congenital anomaly, but who's counting? A national survey of congenital anomalies surveillance in Canada. Chronic Dis Can 27: 36-38, 2006.
4. Lowry RB, Kohut R, Sibbald B & Rouleau J. Anophthalmia and microphthalmia in the Alberta Congenital Anomalies Surveillance System. Can J Ophthalmol 40:38-44, 2005.
5. Lowry RB, Sibbald B, Bamforth JS Re: An epidemiologic analysis of CHARGE Syndrome: preliminary results from a Canadian study (letter). Am J Med Gen 139A: 169, 2005.
6. Wang FL, Gabos S, Sibbald B, Lowry RB Completeness and accuracy of the birth registry data on congenital anomalies in Alberta, Canada Chronic Diseases in Canada 2001; 22(2): 57-66
7. Articles for Canadian Congenital Anomalies Surveillance System Current Contents (http://www.phac-aspc.gc.ca/ccasn-rcsac/index.html):
    i. Sibbald B and Lowry RB Orofacial clefts in Alberta 1980-2004 inclusive (winter 2005)
    http://www.phac-aspc.gc.ca/ccasn-rcsac/ct2005/or-cl-alberta_e.html
    ii. Sibbald B and Lowry RB Abdominal wall defects- Alberta 1980-2002 (winter 2004)
    http://www.phac-aspc.gc.ca/ccasn-rcsac/ct2004/awd-alb.html
    iii. Sibbald B and Lowry RB Down Syndrome in Alberta: Alberta Congenital Anomalies Surveillance System (fall 2003)
    http://www.phac-aspc.gc.ca/ccasn-rcsac/ct2003/abds_e.html
8. Lowry RB. Maternal Ethnicity and risk of Neural Tube Defects. CMAJ 172: 159-160, 2005.
9. Banhidy F, Lowry RB, Czeizel AE. Risk and benefit of drug use during pregnancy. Int J Med Sci 2:100-106, 2005
10. Botto LD & 18 others including Lowry RB. Trends of Selected Malformations in Relation to Folic Acid Recommendations and Fortification: An International Assessment. Birth Defects Research (Part A) 76: 693-705, 2006
11. Lowry RB. The fetal alert network. J Obstet Gynaecol Can 29: 307, 2007
12. Mastroiacovo P and 27 others including Lowry RB. Gastroschisis and Associated Defects: An International Study. Am J Med Genet 143A: 660-671, 2007
13. Lowry RB. Prevalence of anorectal malformations. Orphanet Journal of Rare Diseases 2: 33doi:1186/1750-1172-2-33, 2007.
14. Lowry RB, Sibbald B. The Fetal Alert Network: surveying congenital anomalies. Paediatr Child health 12:713, 2007.
15. De Wals P, Van Allen MI, Lowry RB, Evans JA, Van den Hof MC, Crowley M, Tairou F, Uh SH, Sibbald B, Zimmer P, Fernandez B, Lee NS, Niyonsenga T. Impact of folic acid food fortification on the birth prevalence of lipomyelomeningocele in Canada. Birth defects research (Part A) 82: 106-109, 2008.

16. Lowry RB. Congenital Anomalies Surveillance in Canada. Can J Publ Health 99 : 483-485, 2008.
17. Godwin KA, Kuzeljevic MA, Sibbald B, Lowry RB, Bedard T, Arbour L. Changes in Frequencies of Select Congenital Anomalies since the Onset of Folic Acid Fortification in a Canadian Birth Defect registry. Can J Publ Health 99 : 271-275, 2008.
18. Leoncini E & 28 others including Lowry RB. Frequency of Holoprosencephaly in the International Clearinghouse Birth Defects Surveillance Systems: Searching for Population Variations. Birth Defects Research (Part A) 82 : 585-591, 2008.
19. Mastroiacovo P et al. The Incidence of Gastroschisis ; Research Urgently Needs Resources. BMJ 332 : 423-424, 2006.

ALBERTA CONGENITAL ANOMALIES SURVEILLANCE SYSTEM

6. APPENDICES

Appendix A.1 Flowchart of the Process of ACASS Data Collection

Appendix A.2 Congenital Anomaly(ies) Reporting Form (CARF)

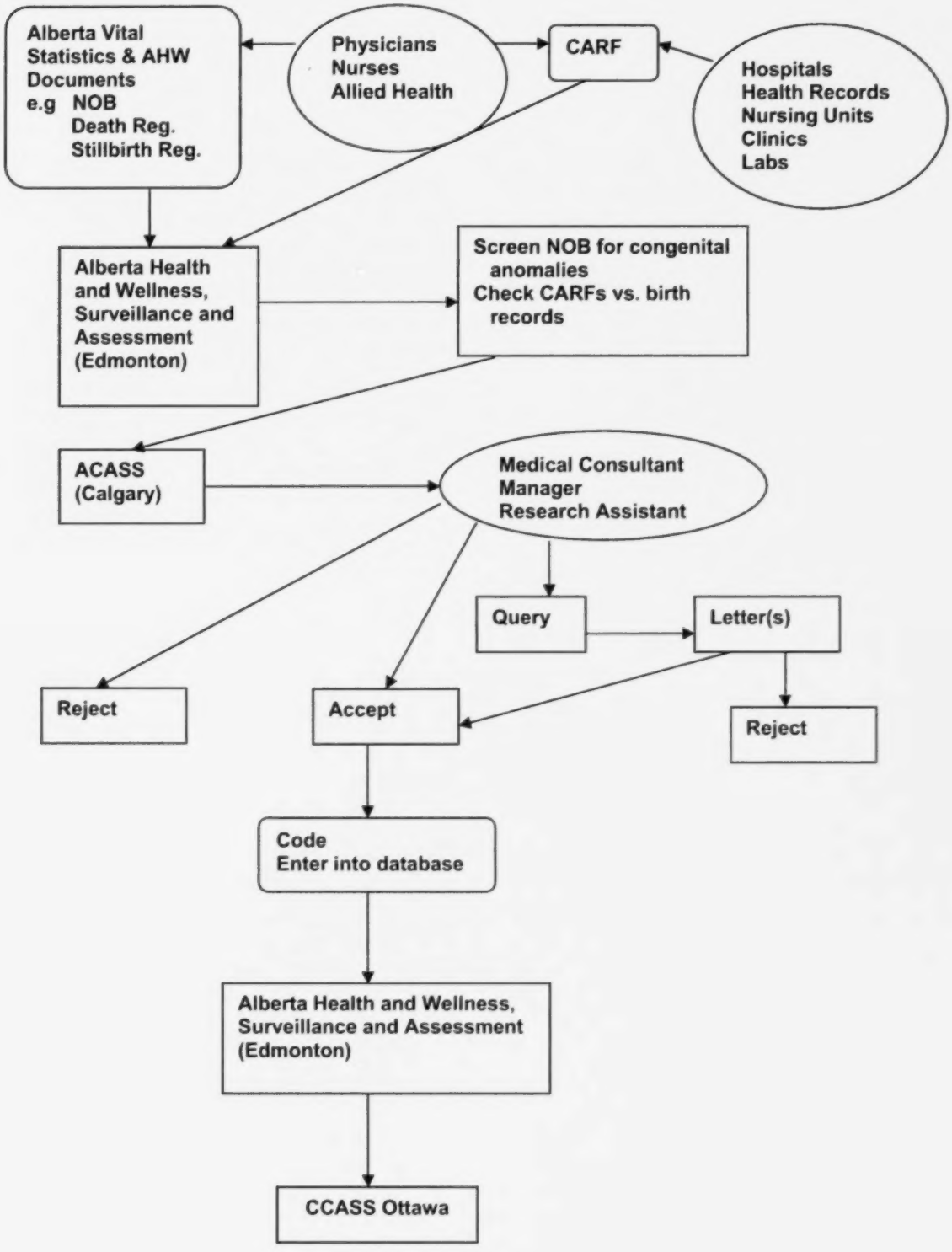
Appendix A.3 Selected Anomaly Rates per 1,000 Total Births

Appendix A.4 Selected anomalies with rates of live births and stillbirths compared with total rates including terminations of pregnancy (ToP)

Appendix A.5 Numbers of cases, anomalies and anomalies per case 1980-2004

Appendix A.6 Termination of pregnancy (ToP) for Congenital anomalies

Appendix A.1 Flowchart of the Process of ACASS Data Collection



Appendix A.2 Congenital Anomaly(ies) Reporting Form (CARF)

Alberta Health and Wellness

Death Reg No | Birth Reg No

Addressograph

Mail parts one and two to:
Alberta Health and Wellness
Surveillance and Environmental Health Branch
PO Box 1360 Stn Main
Edmonton AB T5J 2N3

Congenital Anomaly(ies) Reporting

Fetus / Infant

PLEASE PRINT CLEARLY

Name (*Last, First, Initial*) | Date of Birth (Month by Name / Day / Year)

Gender	Type of Birth	Name of Hospital of Birth
☐ Male ☐ Female ☐ Unknown	☐ Livebirth ☐ Stillbirth ☐ Fetus less than 20 weeks gestation	
Birthweight (Grams)	Gestation Age (Completed Weeks)	Location of Hospital of Birth (City/Town)
Child's Personal Health Number		Attending Physician's Name
Plurality of Birth: ☐ Single ☐ Twin ☐ First ☐ Second ☐ Triplets ☐ First ☐ Second ☐ Third		Physician Responsible for Ongoing Care (*if different from above*)

Parents

Mother's Name (*Last, First, Maiden*)	Date of Birth or Age (*if DOB unavailable*) (Month by Name / Day / Year)	Total Number of: Livebirths
Permanent Address	Mother's Personal Health Number	Stillbirths
City/Town	Postal Code	Spontaneous Abortions
Father's Name (*Last, First, Initial*)	Date of Birth or Age (*if DOB unavailable*) (Month by Name / Day / Year)	Therapeutic Abortions

Reporting Hospital/Agency/Clinic

Name	Infant's Admission (*if different from birthdate*) (Month by Name / Day / Year)	Infant's Discharge (Month by Name / Day / Year)
Location (*City/Town*)		Infant's Death (If Applicable) (Month by Name / Day / Year)

Full description of Congenital Anomaly(ies) and/or **SYNDROME DIAGNOSES** (*If necessary, please attach supporting documents.*)

OFFICE USE ONLY

Completed by	Position	Date

HS0020-112 (2008/06)

Send to Surveillance and Environmental Health

Appendix A.3 **Alberta Congenital Anomalies Surveillance System**

Live Birth and Stillbirth Single Year Anomaly Rates per 1,000 Total Births

Diagnostic Category		2003	2004	2005	2006	2007
Anencephaly	NUMBER	9	3	7	7	7
	RATE	**0.22**	**0.07**	**0.17**	**0.16**	**0.14**
ICD-9 7400.., 7401..	Lower CI	0.10	0.01	0.07	0.06	0.06
ICD-10 Q00.00, Q00.01, Q00.1	Upper CI	0.42	0.21	0.34	0.32	0.29
Spina Bifida without Anencephaly	NUMBER	11	14	8	25	16
	RATE	**0.27**	**0.35**	**0.19**	**0.56**	**0.33**
ICD-9 741..	Lower CI	0.14	0.19	0.08	0.36	0.19
ICD-10 Q05..	Upper CI	0.49	0.58	0.37	0.82	0.53
Encephalocele	NUMBER	3	3	6	2	3
	RATE	**0.07**	**0.07**	**0.14**	**0.04**	**0.06**
ICD-9 7420..	Lower CI	0.02	0.01	0.05	0.01	0.01
ICD-10 Q01..	Upper CI	0.21	0.21	0.31	0.15	0.17
Neural Tube Defects (all)	NUMBER	23	20	21	34	26
	RATE	**0.57**	**0.49**	**0.50**	**0.76**	**0.53**
ICD-9 740.., 741.., 742..	Lower CI	0.36	0.30	0.31	0.52	0.35
ICD-10 Q00.., Q01.., Q05..	Upper CI	0.86	0.76	0.77	1.06	0.78
Hydrocephalus without Spina Bifida (Excl. hydranencephaly)	NUMBER	22	21	21	36	29
	RATE	**0.55**	**0.52**	**0.50**	**0.80**	**0.60**
ICD-9 7423.., excl. 74232	Lower CI	0.34	0.32	0.31	0.56	0.40
ICD-10 Q03	Upper CI	0.83	0.79	0.77	1.11	0.86
Microcephaly	NUMBER	20	11	23	13	13
	RATE	**0.50**	**0.27**	**0.55**	**0.29**	**0.27**
ICD-9 7421..	Lower CI	0.30	0.14	0.35	0.15	0.14
ICD-10 Q02	Upper CI	0.77	0.48	0.82	0.49	0.46
Anophthalmia/microphthalmia	NUMBER	2	8	14	2	7
	RATE	**0.05**	**0.20**	**0.33**	**0.04**	**0.14**
ICD-9 7430.., 7431..	Lower CI	0.01	0.09	0.18	0.01	0.06
ICD-10 Q11.0, Q11.1, Q11.2	Upper CI	0.17	0.39	0.56	0.15	0.29
Congenital cataract	NUMBER	4	4	5	4	9
	RATE	**0.10**	**0.10**	**0.12**	**0.09**	**0.18**
ICD-9 7432..	Lower CI	0.03	0.03	0.04	0.02	0.08
ICD-10 Q12.0	Upper CI	0.25	0.25	0.27	0.22	0.35
Aniridia	NUMBER	2	1	0	0	0
	RATE	**0.05**	**0.02**	**0**	**0**	**0**
ICD-9 74342	Lower CI	0.01	0.00			
ICD-10 Q13.1	Upper CI	0.17	0.12			
Anotia/microtia	NUMBER	7	13	12	8	13
	RATE	**0.17**	**0.32**	**0.29**	**0.18**	**0.27**
ICD-9 74401, 74421	Lower CI	0.07	0.17	0.15	0.08	0.14
ICD-10 Q16.0, Q17.2	Upper CI	0.36	0.55	0.50	0.35	0.46

Number = Defects occurring in Live Births and Stillbirths ≥ 20 weeks or ≥ 500 g CI = Approximate 95% Confidence Intervals

Appendix A.3 Continued **Alberta Congenital Anomalies Surveillance System**

Live Birth and Stillbirth Aggregate Year Anomaly Rates per 1,000 Total Births

Diagnostic Category		80-89 (10 years)	90-99 (10 years)	00-04 (5 years)	05-07 (3 years)
Anencephaly	NUMBER	148	75	26	21
	RATE	**0.33**	**0.19**	**0.13**	**0.15**
ICD-9 7400.., 7401..	Lower CI	0.28	0.15	0.09	0.10
ICD-10 Q00.00, Q00.01, Q00.1	Upper CI	0.39	0.24	0.20	0.24
Spina Bifida without Anencephaly	NUMBER	218	198	48	49
	RATE	**0.51**	**0.49**	**0.25**	**0.36**
ICD-9 741..	Lower CI	0.44	0.43	0.18	0.27
ICD-10 Q05..	Upper CI	0.58	0.57	0.33	0.48
Encephalocele	NUMBER	42	36	24	11
	RATE	**0.10**	**0.09**	**0.12**	**0.08**
ICD-9 7420..	Lower CI	0.07	0.06	0.08	0.04
ICD-10 Q01..	Upper CI	0.13	0.13	0.18	0.14
Neural Tube Defects (all)	NUMBER	407	307	98	81
	RATE	**0.95**	**0.77**	**0.51**	**0.60**
ICD-9 740.., 741.., 742..	Lower CI	0.86	0.69	0.41	0.47
ICD-10 Q00.., Q01.., Q05..	Upper CI	1.04	0.87	0.62	0.74
Hydrocephalus without Spina Bifida (Excl. hydranencephaly)	NUMBER	221	180	104	86
	RATE	**0.51**	**0.45**	**0.54**	**0.63**
ICD-9 7423.., excl. 74232	Lower CI	0.45	0.39	0.44	0.51
ICD-10 Q03	Upper CI	0.59	0.53	0.65	0.78
Microcephaly	NUMBER	159	123	71	49
	RATE	**0.41**	**0.32**	**0.37**	**0.36**
ICD-9 7421..	Lower CI	0.33	0.26	0.29	0.27
ICD-10 Q02	Upper CI	0.50	0.38	0.46	0.48
Anophthalmia/microphthalmia	NUMBER	53	66	29	23
	RATE	**0.12**	**0.17**	**0.15**	**0.17**
ICD-9 7430.., 7431..	Lower CI	0.09	0.13	0.10	0.11
ICD-10 Q11.0, Q11.1, Q11.2	Upper CI	0.16	0.21	0.22	0.25
Congenital cataract	NUMBER	62	54	16	18
	RATE	**0.14**	**0.14**	**0.08**	**0.13**
ICD-9 7432..	Lower CI	0.11	0.10	0.05	0.08
ICD-10 Q12.0	Upper CI	0.18	0.18	0.13	0.21
Aniridia	NUMBER	0	4	3	0
	RATE	**0**	**0.01**	**0.02**	**0**
ICD-9 74342	Lower CI		0.00	0.00	
ICD-10 Q13.1	Upper CI		0.03	0.04	
Anotia/microtia	NUMBER	27	58	39	33
	RATE	**0.06**	**0.15**	**0.20**	**0.24**
ICD-9 74401, 74421	Lower CI	0.04	0.11	0.14	0.17
ICD-10 Q16.0, Q17.2	Upper CI	0.09	0.19	0.28	0.34

Number = Defects occurring in Live Births and Stillbirths ≥ 20 weeks or ≥ 500 g CI = Approximate 95% Confidence Intervals

Live Birth and Stillbirth Single Year Anomaly Rates per 1,000 Total Births

Diagnostic Category		2003	2004	2005	2006	2007
Common Truncus	NUMBER	2	1	2	0	5
Excludes AP window	**RATE**	**0.05**	**0.02**	**0.05**	**0**	**0.10**
ICD-9 74500	Lower CI	0.01	0.00	0.01		0.03
ICD-10 Q20.0	Upper CI	0.17	0.12	0.16		0.24
Transposition of Great Arteries	NUMBER	17	18	19	15	13
	RATE	**0.42**	**0.44**	**0.45**	**0.33**	**0.27**
ICD-9 7451..	Lower CI	0.25	0.26	0.27	0.19	0.14
ICD-10 Q20.11, Q20.3, Q20.5	Upper CI	0.68	0.70	0.71	0.55	0.45
Tetralogy of Fallot	NUMBER	10	12	17	17	20
	RATE	**0.25**	**0.30**	**0.41**	**0.38**	**0.41**
ICD-9 74520 - 74523	Lower CI	0.12	0.15	0.24	0.22	0.25
ICD-10 Q21.3.., Q21.82	Upper CI	0.46	0.51	0.65	0.60	0.63
Ventricular Septal Defect	NUMBER	112	156	144	118	133
	RATE	**2.79**	**3.85**	**3.44**	**2.62**	**2.73**
ICD-9 7454..	Lower CI	2.30	3.27	2.90	2.17	2.29
ICD-10 Q21.0	Upper CI	3.36	4.50	4.05	3.15	3.24
Atrial Septal Defect	NUMBER	78	94	86	61	69
	RATE	**1.94**	**2.32**	**2.05**	**1.36**	**1.42**
ICD-9 7455..	Lower CI	1.54	1.87	1.64	1.04	1.10
ICD-10 Q21.1..	Upper CI	2.43	2.84	2.54	1.74	1.79
Endocardial Cushion Defect	NUMBER	19	24	15	12	25
	RATE	**0.47**	**0.59**	**0.36**	**0.27**	**0.51**
ICD-9 7456..	Lower CI	0.29	0.38	0.20	0.14	0.33
ICD-10 Q21.2..	Upper CI	0.74	0.88	0.59	0.46	0.76
Pulmonary Valve Atresia and Stenosis	NUMBER	23	18	26	20	32
	RATE	**0.57**	**0.44**	**0.62**	**0.44**	**0.66**
ICD-9 74600, 74601	Lower CI	0.36	0.26	0.41	0.27	0.45
ICD-10 Q22.0, Q22.1	Upper CI	0.86	0.70	0.91	0.69	0.93
Tricuspid Valve Atresia and Stenosis	NUMBER	4	3	1	4	7
	RATE	**0.10**	**0.07**	**0.02**	**0.09**	**0.14**
ICD-9 74610	Lower CI	0.03	0.01	0.00	0.02	0.06
ICD-10 Q22.4	Upper CI	0.25	0.21	0.12	0.22	0.29
Ebstein's Anomaly	NUMBER	2	1	1	4	2
	RATE	**0.05**	**0.02**	**0.02**	**0.09**	**0.04**
ICD-9 74620	Lower CI	0.01	0.00	0.00	0.02	0.00
ICD-10 Q22.5	Upper CI	0.17	0.12	0.12	0.22	0.14
Aortic Valve Stenosis	NUMBER	8	11	4	4	11
	RATE	**0.20**	**0.27**	**0.10**	**0.09**	**0.23**
ICD-9 74630	Lower CI	0.09	0.14	0.03	0.02	0.11
ICD-10 Q23.0	Upper CI	0.39	0.48	0.24	0.22	0.40

Number = Defects occurring in Live Births and Stillbirths ≥ 20 weeks or ≥ 500 g CI = Approximate 95% Confidence Intervals

Live Birth and Stillbirth Aggregate Year Anomaly Rates per 1,000 Total Births

Diagnostic Category		80-89 (10 years)	90-99 (10 years)	00-04 (5 years)	05-07 (3 years)
Common Truncus	NUMBER	31	22	7	7
Excludes AP window	**RATE**	**0.07**	**0.06**	**0.04**	**0.05**
ICD-9 74500	Lower CI	0.05	0.03	0.01	0.02
ICD-10 Q20.0	Upper CI	0.10	0.08	0.07	0.11
Transposition of Great Arteries	NUMBER	128	127	77	47
	RATE	**0.30**	**0.32**	**0.40**	**0.35**
ICD-9 7451..	Lower CI	0.25	0.27	0.31	0.26
ICD-10 Q20.11, Q20.3, Q20.5	Upper CI	0.35	0.38	0.50	0.46
Tetralogy of Fallot	NUMBER	85	107	57	54
	RATE	**0.20**	**0.27**	**0.29**	**0.40**
ICD-9 74520 - 74523	Lower CI	0.16	0.22	0.22	0.30
ICD-10 Q21.3.., Q21.82	Upper CI	0.24	0.33	0.38	0.52
Ventricular Septal Defect	NUMBER	1185	1130	601	395
	RATE	**2.75**	**2.85**	**3.11**	**2.91**
ICD-9 7454..	Lower CI	2.60	2.69	2.86	2.63
ICD-10 Q21.0	Upper CI	2.91	3.02	3.36	2.22
Atrial Septal Defect	NUMBER	492	726	353	216
	RATE	**1.14**	**1.83**	**1.82**	**1.59**
ICD-9 7455..	Lower CI	1.04	1.70	1.64	1.39
ICD-10 Q21.1..	Upper CI	1.25	1.97	2.02	1.82
Endocardial Cushion Defect	NUMBER	159	151	92	52
	RATE	**0.37**	**0.38**	**0.48**	**0.38**
ICD-9 7456..	Lower CI	0.31	0.32	0.38	0.29
ICD-10 Q21.2..	Upper CI	0.43	0.45	0.58	0.50
Pulmonary Valve Atresia and Stenosis	NUMBER	327	245	104	78
	RATE	**0.76**	**0.62**	**0.54**	**0.58**
ICD-9 74600, 74601	Lower CI	0.68	0.54	0.44	0.46
ICD-10 Q22.0, Q22.1	Upper CI	0.85	0.70	0.65	0.72
Tricuspid Valve Atresia and Stenosis	NUMBER	41	31	12	12
	RATE	**0.10**	**0.08**	**0.06**	**0.09**
ICD-9 74610	Lower CI	0.07	0.05	0.03	0.05
ICD-10 Q22.4	Upper CI	0.13	0.11	0.11	0.15
Ebstein's Anomaly	NUMBER	20	21	12	7
	RATE	**0.05**	**0.05**	**0.06**	**0.05**
ICD-9 74620	Lower CI	0.03	0.03	0.03	0.02
ICD-10 Q22.5	Upper CI	0.07	0.08	0.11	0.11
Aortic Valve Stenosis	NUMBER	60	71	36	19
	RATE	**0.14**	**0.18**	**0.19**	**0.14**
ICD-9 74630	Lower CI	0.11	0.14	0.13	0.08
ICD-10 Q23.0	Upper CI	0.18	0.23	0.26	0.22

Number = Defects occurring in Live Births and Stillbirths ≥ 20 weeks or ≥ 500 g CI = Approximate 95% Confidence Intervals

Live Birth and Stillbirth Single Year Anomaly Rates per 1,000 Total Births

Diagnostic Category		2003	2004	2005	2006	2007
Hypoplastic Left Heart Syndrome	NUMBER	7	10	19	13	17
	RATE	**0.17**	**0.25**	**0.45**	**0.29**	**0.35**
ICD-9 74670	Lower CI	0.07	0.12	0.27	0.15	0.20
ICD-10 Q23.4	Upper CI	0.36	0.45	0.71	0.49	0.56
Patent Ductus Arteriosus	NUMBER	25	32	34	21	37
	RATE	**0.62**	**0.79**	**0.81**	**0.47**	**0.76**
ICD-9 74700	Lower CI	0.40	0.54	0.56	0.29	0.54
ICD-10 Q25.0	Upper CI	0.92	1.11	1.13	0.71	1.05
Coarctation of the Aorta	NUMBER	13	13	16	12	19
	RATE	**0.32**	**0.32**	**0.38**	**0.27**	**0.39**
ICD-9 7471..	Lower CI	0.17	0.17	0.22	0.14	0.24
ICD-10 Q25.1..	Upper CI	0.55	0.55	0.62	0.46	0.61
Cleft Palate without Cleft Lip	NUMBER	24	33	29	25	25
	RATE	**0.60**	**0.81**	**0.69**	**0.56**	**0.51**
ICD-9 7490..	Lower CI	0.38	0.56	0.46	0.36	0.33
ICD-10 Q35..	Upper CI	0.89	1.14	0.99	0.82	0.76
Cleft Lip with and without Cleft Palate	NUMBER	45	47	45	42	79
	RATE	**1.12**	**1.16**	**1.09**	**0.93**	**1.62**
ICD-9 7491.., 7492..	Lower CI	0.82	0.85	0.78	0.67	1.29
ICD-10 Q36.., Q37..	Upper CI	1.50	1.54	1.44	1.26	2.02
Choanal Atresia	NUMBER	6	9	7	8	5
	RATE	**0.15**	**0.22**	**0.17**	**0.18**	**0.10**
ICD-9 7480..	Lower CI	0.05	0.10	0.07	0.08	0.03
ICD-10 Q03.0..	Upper CI	0.32	0.42	0.34	0.35	0.24
Oesophageal Atresia/ Tracheo-oesphageal Fistula	NUMBER	7	12	8	12	5
	RATE	**0.17**	**0.30**	**0.19**	**0.27**	**0.10**
ICD-9 75030-75035	Lower CI	0.07	0.15	0.08	0.14	0.03
ICD-10 Q39.0 – Q39.4	Upper CI	0.36	0.51	0.37	0.46	0.24
Rectal and Large Intestinal Atresia/Stenosis	NUMBER	25	27	18	22	11
	RATE	**0.62**	**0.67**	**0.43**	**0.49**	**0.23**
ICD-9 7512..	Lower CI	0.40	0.44	0.26	0.31	0.11
ICD-10 Q42..	Upper CI	0.92	0.97	0.68	0.74	0.40
Pyloric Stenosis	NUMBER	40	33	42	43	51
	RATE	**1.00**	**0.81**	**1.00**	**0.96**	**1.05**
ICD-9 75051	Lower CI	0.71	0.56	0.72	0.69	0.78
ICD-10 Q40.0	Upper CI	1.36	1.14	1.36	1.29	1.38
Hirschsprung Disease	NUMBER	8	5	5	12	6
	RATE	**0.20**	**0.12**	**0.12**	**0.27**	**0.12**
ICD-9 7513..	Lower CI	0.09	0.04	0.04	0.14	0.05
ICD-10 Q43.1..	Upper CI	0.39	0.28	0.27	0.46	0.27

Number = Defects occurring in Live Births and Stillbirths ≥ 20 weeks or ≥ 500 g CI = Approximate 95% Confidence Intervals

Live Birth and Stillbirth Aggregate Year Anomaly Rates per 1,000 Total Births

Diagnostic Category		80-89 (10 years)	90-99 (10 years)	00-04 (5 years)	05-07 (3 years)
Hypoplastic Left Heart Syndrome	NUMBER	93	90	50	49
	RATE	**0.22**	**0.23**	**0.26**	**0.36**
ICD-9 74670	Lower CI	0.17	0.18	0.19	0.27
ICD-10 Q23.4	Upper CI	0.26	0.28	0.34	0.48
Patent Ductus Arteriosus	NUMBER	919	317	122	92
	RATE	**2.13**	**0.80**	**0.63**	**0.68**
ICD-9 74700	Lower CI	2.00	0.71	0.52	0.55
ICD-10 Q25.0	Upper CI	2.28	0.89	0.75	0.83
Coarctation of the Aorta	NUMBER	171	177	60	47
	RATE	**0.40**	**0.45**	**0.31**	**0.35**
ICD-9 7471..	Lower CI	0.34	0.38	0.24	0.26
ICD-10 Q25.1..	Upper CI	0.46	0.52	0.40	0.46
Cleft Palate without Cleft Lip	NUMBER	282	332	156	79
	RATE	**0.65**	**0.84**	**0.81**	**0.58**
ICD-9 7490..	Lower CI	0.58	0.75	0.68	0.46
ICD-10 Q35..	Upper CI	0.74	0.93	0.94	0.73
Cleft Lip with and without Cleft Palate	NUMBER	479	467	228	166
	RATE	**1.11**	**1.18**	**1.18**	**1.22**
ICD-9 7491.., 7492..	Lower CI	1.02	1.07	1.03	1.05
ICD-10 Q36.., Q37..	Upper CI	1.22	1.29	1.34	1.43
Choanal Atresia	NUMBER	59	61	39	20
	RATE	**0.14**	**0.15**	**0.20**	**0.15**
ICD-9 7480..	Lower CI	0.10	0.12	0.14	0.09
ICD-10 Q03.0..	Upper CI	0.18	0.20	0.28	0.33
Oesophageal Atresia/ Tracheo-oesphageal Fistula	NUMBER	129	96	39	25
	RATE	**0.30**	**0.24**	**0.20**	**0.18**
ICD-9 75030-75035	Lower CI	0.25	0.20	0.14	0.12
ICD-10 Q39.0 – Q39.4	Upper CI	0.36	0.30	0.28	0.27
Rectal and Large Intestinal Atresia/Stenosis	NUMBER	205	224	124	51
	RATE	**0.48**	**0.57**	**0.64**	**0.38**
ICD-9 7512..	Lower CI	0.41	0.49	0.53	0.28
ICD-10 Q42..	Upper CI	0.55	0.64	0.76	0.50
Pyloric Stenosis	NUMBER	386	281	179	136
	RATE	**0.90**	**0.71**	**0.92**	**1.00**
ICD-9 75051	Lower CI	0.81	0.63	0.79	0.84
ICD-10 Q40.0	Upper CI	0.99	0.80	1.07	1.19
Hirschsprung Disease	NUMBER	62	56	22	23
	RATE	**0.14**	**0.14**	**0.11**	**0.17**
ICD-9 7513..	Lower CI	0.11	0.11	0.07	0.11
ICD-10 Q43.1..	Upper CI	0.18	0.18	0.17	0.25

Number = Defects occurring in Live Births and Stillbirths ≥ 20 weeks or ≥ 500 g CI = Approximate 95% Confidence Intervals

Live Birth and Stillbirth Single Year Anomaly Rates per 1,000 Total Births

Diagnostic Category		2003	2004	2005	2006	2007
Biliary Atresia	NUMBER	2	5	5	1	3
	RATE	**0.05**	**0.12**	**0.12**	**0.02**	**0.06**
ICD-9 75165	Lower CI	0.01	0.04	0.04	0.00	0.01
ICD-10 Q44.2	Upper CI	0.17	0.28	0.27	0.11	0.17
Renal Agenesis/Hypoplasia	NUMBER	18	20	17	19	27
	RATE	**0.45**	**0.49**	**0.41**	**0.42**	**0.55**
ICD-9 7530..	Lower CI	0.27	0.30	0.24	0.25	0.37
ICD-10 Q60..	Upper CI	0.71	0.76	0.65	0.66	0.81
Bladder Exstrophy	NUMBER	2	1	1	1	2
	RATE	**0.05**	**0.02**	**0.02**	**0.02**	**0.04**
ICD-9 75350	Lower CI	0.01	0.00	0.00	0.00	0.00
ICD-10 Q64.1 (excl Q64.10)	Upper CI	0.17	0.12	0.12	0.11	0.14
Obstructive Genitourinary	NUMBER	74	73	73	74	89
Defect	**RATE**	**1.84**	**1.80**	**1.74**	**1.65**	**1.83**
ICD-9 7532.., 7536..	Lower CI	1.45	1.41	1.37	1.29	1.47
ICD-10 Q62.0 – Q62.3, Q64.2, Q64.3	Upper CI	2.31	2.26	2.19	2.07	2.25
Hypospadias/Epispadias	NUMBER	89	95	100	81	107
denominator male births only	**RATE**	**4.33**	**4.56**	**4.67**	**3.52**	**4.32**
ICD-9 75260, 75261	Lower CI	3.48	3.69	3.80	2.80	3.54
ICD-10 Q54 (excl. Q54.4), Q64.0	Upper CI	5.33	5.58	5.68	4.37	5.22
Reduction Deformity, Upper	NUMBER	30	27	23	16	27
Limbs	**RATE**	**0.75**	**0.67**	**0.55**	**0.36**	**0.55**
ICD-9 7552..	Lower CI	0.51	0.44	0.35	0.20	0.37
ICD-10 Q71..	Upper CI	1.07	0.97	0.82	0.58	0.81
Reduction Deformity, Lower	NUMBER	11	13	12	13	18
Limbs	**RATE**	**0.27**	**0.32**	**0.29**	**0.29**	**0.37**
ICD-9 7553..	Lower CI	0.14	0.17	0.15	0.15	0.22
ICD-10 Q72..	Upper CI	0.49	0.55	0.50	0.49	0.58
Gastroschisis	NUMBER	13	13	22	27	24
	RATE	**0.32**	**0.32**	**0.53**	**0.60**	**0.49**
ICD-9 75671	Lower CI	0.17	0.17	0.33	0.40	0.32
ICD-10 Q79.3	Upper CI	0.55	0.55	0.79	0.87	0.73
Omphalocele	NUMBER	8	9	4	7	9
	RATE	**0.20**	**0.22**	**0.10**	**0.16**	**0.18**
ICD-9 75670	Lower CI	0.09	0.10	0.03	0.06	0.08
ICD-10 Q79.2	Upper CI	0.39	0.42	0.24	0.32	0.35
Congenital Hip Dislocation	NUMBER	15	25	22	24	15
	RATE	**0.37**	**0.62**	**0.53**	**0.53**	**0.31**
ICD-9 75430	Lower CI	0.21	0.40	0.33	0.34	0.17
ICD-10 Q65.0-Q65.2 (excl. dysplasia)	Upper CI	0.62	0.91	0.79	0.79	0.51

Number = Defects occurring in Live Births and Stillbirths ≥ 20 weeks or ≥ 500 g CI = Approximate 95% Confidence Intervals

Appendix A.3 Continued **Alberta Congenital Anomalies Surveillance System**

Live Birth and Stillbirth Aggregate Year Anomaly Rates per 1,000 Total Births

Diagnostic Category		80-89 (10 years)	90-99 (10 years)	00-04 (5 years)	05-07 (3 years)
Biliary Atresia	NUMBER	20	24	12	9
	RATE	**0.05**	**0.06**	**0.06**	**0.07**
ICD-9 75165	Lower CI	0.03	0.04	0.03	0.03
ICD-10 Q44.2	Upper CI	0.07	0.09	0.11	0.13
Renal Agenesis/Hypoplasia	NUMBER	167	179	101	63
	RATE	**0.39**	**0.45**	**0.52**	**0.46**
ICD-9 7530..	Lower CI	0.33	0.39	0.43	0.36
ICD-10 Q60..	Upper CI	0.45	0.52	0.63	0.59
Bladder Exstrophy	NUMBER	12	11	10	4
	RATE	**0.03**	**0.03**	**0.05**	**0.03**
ICD-9 75350	Lower CI	0.01	0.01	0.02	0.01
ICD-10 Q64.1 (excl Q64.10)	Upper CI	0.05	0.05	0.09	0.07
Obstructive Genitourinary Defect	NUMBER	405	609	359	236
	RATE	**0.94**	**1.54**	**1.85**	**1.74**
ICD-9 7532.., 7536..	Lower CI	0.85	1.42	1.67	1.53
ICD-10 Q62.0 – Q62.3, Q64.2, Q64.3	Upper CI	1.04	1.66	2.06	1.98
Hypospadias/Epispadias	NUMBER	911	851	413	288
denominator male births only	**RATE**	**4.13**	**4.19**	**4.17**	**4.16**
ICD-9 75260, 75261	Lower CI	3.86	3.91	3.78	3.70
ICD-10 Q54 (excl. Q54.4), Q64.0	Upper CI	4.40	4.48	4.59	4.67
Reduction Deformity, Upper Limbs	NUMBER	224	264	127	66
	RATE	**0.52**	**0.67**	**0.66**	**0.49**
ICD-9 7552..	Lower CI	0.45	0.59	0.55	0.38
ICD-10 Q71..	Upper CI	0.59	0.75	0.78	0.62
Reduction Deformity, Lower Limbs	NUMBER	336	284	72	43
	RATE	**0.78**	**0.72**	**0.37**	**0.32**
ICD-9 7553..	Lower CI	0.70	0.64	0.29	0.23
ICD-10 Q72..	Upper CI	0.87	0.80	0.47	0.43
Gastroschisis	NUMBER	61	79	61	73
	RATE	**0.14**	**0.20**	**0.32**	**0.54**
ICD-9 75671	Lower CI	0.11	0.16	0.24	0.42
ICD-10 Q79.3	Upper CI	0.18	0.25	0.40	0.68
Omphalocele	NUMBER	88	79	40	20
	RATE	**0.20**	**0.20**	**0.21**	**0.15**
ICD-9 75670	Lower CI	0.16	0.16	0.15	0.09
ICD-10 Q79.2	Upper CI	0.25	0.25	0.28	0.33
Congenital Hip Dislocation	NUMBER	619	491	128	61
	RATE	**1.44**	**1.24**	**0.66**	**0.45**
ICD-9 75430	Lower CI	1.33	1.13	0.55	0.34
ICD-10 Q65.0-Q65.2 (excl. dysplasia)	Upper CI	1.56	1.35	0.79	0.58

Number = Defects occurring in Live Births and Stillbirths ≥ 20 weeks or ≥ 500 g CI = Approximate 95% Confidence Intervals

Appendix A.3 Continued **Alberta Congenital Anomalies Surveillance System**

Live Birth and Stillbirth Single Year Anomaly Rates per 1,000 Total Births

Diagnostic Category		2003	2004	2005	2006	2007
Diaphragmatic Hernia	NUMBER	12	5	15	10	12
	RATE	**0.30**	**0.12**	**0.36**	**0.22**	**0.25**
ICD-9 75660, 75661	Lower CI	0.15	0.04	0.20	0.11	0.13
ICD-10 Q79.0.., Q79.11, Q79.12	Upper CI	0.52	0.28	0.59	0.41	0.43
Trisomy 13	NUMBER	5	7	4	8	12
	RATE	**0.12**	**0.17**	**0.10**	**0.18**	**0.25**
ICD-9 7581..	Lower CI	0.04	0.07	0.03	0.08	0.13
ICD-10 Q91.4-Q91.7	Upper CI	0.29	0.35	0.24	0.35	0.43
Down Syndrome (Trisomy 21)	NUMBER	72	68	86	60	81
	RATE	**1.79**	**1.68**	**2.05**	**1.33**	**1.66**
ICD-9 7580..	Lower CI	1.41	1.30	1.64	1.02	1.32
ICD-10 Q90..	Upper CI	2.26	2.12	2.53	1.72	2.07
Trisomy 18	NUMBER	5	12	18	12	15
	RATE	**0.12**	**0.30**	**0.43**	**0.27**	**0.31**
ICD-9 7582..	Lower CI	0.04	0.15	0.26	0.14	0.17
ICD-10 Q91.0-Q91.3	Upper CI	0.29	0.51	0.68	0.46	0.51

Number = Defects occurring in Live Births and Stillbirths ≥ 20 weeks or ≥ 500 g CI = Approximate 95% Confidence Intervals

Live Birth and Stillbirth Aggregate Year Anomaly Rates per 1,000 Total Births

Diagnostic Category		80-89 (10 years)	90-99 (10 years)	00-04 (5 years)	05-07 (3 years)
Diaphragmatic Hernia	NUMBER	143	109	67	37
	RATE	**0.33**	**0.27**	**0.35**	**0.27**
ICD-9 75660, 75661	Lower CI	0.28	0.23	0.27	0.19
ICD-10 Q79.0.., Q79.11, Q79.12	Upper CI	0.39	0.33	0.44	0.38
Trisomy 13	NUMBER	32	44	22	24
	RATE	**0.07**	**0.11**	**0.11**	**0.18**
ICD-9 7581..	Lower CI	0.05	0.08	0.07	0.11
ICD-10 Q91.4-Q91.7	Upper CI	0.10	0.15	0.17	0.26
Down Syndrome (Trisomy 21)	NUMBER	402	452	301	227
	RATE	**0.93**	**1.14**	**1.56**	**1.67**
ICD-9 7580..	Lower CI	0.84	1.04	1.38	1.46
ICD-10 Q90..	Upper CI	1.03	1.25	1.74	1.91
Trisomy 18	NUMBER	72	101	57	45
	RATE	**0.17**	**0.25**	**0.29**	**0.33**
ICD-9 7582..	Lower CI	0.13	0.21	0.22	0.24
ICD-10 Q91.0-Q91.3	Upper CI	0.21	0.31	0.38	0.44

Number = Defects occurring in Live Births and Stillbirths ≥ 20 weeks or ≥ 500 g CI = Approximate 95% Confidence Intervals

Appendix A.4 Selected Anomalies with Rates of Live Births (L) and Stillbirths (S) Compared with Total Rates including Terminations of Pregnancy/Fetal Loss (ToP)

Table A.4.1 2000-2004 (5 years)

Congenital Anomalies (CAs)	Number of Anomalies		Rates/1000 Total Births	
	Live (L) and Still (S)	ToP (T)	L + S	L + S + T
Anencephaly	26	22	0.13	0.25
Spina Bifida	48	9	0.25	0.29
Encephalocele	24	4	0.12	0.14
Hydrocephaly	104	8	0.54	0.58
Rectal & Large Intestinal Atresia/Stenosis	124	22	0.64	0.75
Renal Agenesis/Hypoplasia	101	16	0.52	0.60
Limb Reduction Anomalies	199	46	1.03	1.27
Chromosome Anomalies (all)	576	181	2.98	3.91
Down Syndrome	301	77	1.56	1.95
Syndromes (Q87*)	174	4	0.90	0.92

Table A.4.2 2005-2007 (3 years)

Congenital Anomalies (CAs)	Number of Anomalies		Rates/1000 Total Births	
	Live (L) and Still (S)	ToP (T)	L + S	L + S + T
Anencephaly	21	6	0.15	0.20
Spina Bifida	49	9	0.36	0.43
Encephalocele	11	5	0.08	0.12
Hydrocephaly	86	3	0.63	0.66
Rectal & Large Intestinal Atresia/Stenosis	51	6	0.38	0.42
Renal Agenesis/Hypoplasia	63	5	0.46	0.50
Limb Reduction Anomalies	109	23	0.80	0.97
Chromosome Anomalies (all)	441	189	3.25	4.65
Down Syndrome	227	87	1.67	2.32
Syndromes (Q87*)	104	6	0.77	0.81

Appendix A.5 Numbers of Cases, Anomalies and Anomalies per Case 1980-2007, Live Births and Stillbirths

Year	Total Births (Live & Still)	# Cases (Live & Still)	Case Rate/1000 Total Births	# Anomalies (Live & Still)	Anomaly Rate/1000 Total Births	Average # Anomalies/ Case
1980	39655	1366	34.45	1773	44.71	1.30
1981	42463	1444	34.01	1898	44.7	1.31
1982	44987	1570	34.90	2130	47.35	1.36
1983	45401	1486	32.73	2136	47.05	1.44
1984	43856	1583	36.1	2181	49.73	1.38
1985	43565	1688	38.75	2437	55.94	1.44
1986	43555	1778	40.82	2460	56.48	1.38
1987	41956	1694	40.38	2464	58.73	1.45
1988	41967	1872	44.61	2781	66.27	1.49
1989	43218	1929	44.63	2898	67.06	1.50
1990	42892	1985	46.28	3004	70.04	1.51
1991	42670	1779	41.69	2563	60.07	1.44
1992	41942	1777	42.37	2684	63.99	1.51
1993	40159	1472	36.65	2243	55.85	1.52
1994	39718	1409	35.48	2220	55.89	1.58
1995	38782	1166	30.07	1913	49.33	1.64
1996	37707	1137	30.15	1802	47.79	1.58
1997	36797	1080	29.35	1819	49.43	1.68
1998	37715	1144	30.33	1881	49.87	1.64
1999	38044	1172	30.81	2187	57.49	1.87
2000	36860	1258	34.13	2136	57.95	1.70
2001	37454	1347	35.96	2315	61.81	1.72
2002	38540	1334	34.61	2263	58.72	1.70
2003	40120	1448	36.09	2430	60.57	1.68
2004	40570	1505	37.1	2727	67.22	1.81
2005	41890	1547	36.93	2709	64.67	1.75
2006	44954	1535	34.15	2496	55.52	1.63
2007	48683	1763	36.21	2868	58.91	1.63
Total	1156120	42268	36.56	65418	56.58	1.55

Appendix A.6 Termination of Pregnancy or Fetal Loss with Congenital Anomalies, 1997-2007

Year of Termination	# ToP cases	# ToP anomalies	Average # Anomalies Per Case
1997	65	127	1.95
1998	**69**	**229**	**3.32**
1999	54	181	3.35
2000	**52**	**145**	**2.79**
2001	52	177	3.40
2002	**62**	**136**	**2.19**
2003	92	210	2.28
2004	**74**	**201**	**2.72**
2005	92	219	2.38
2006	**103**	**227**	**2.20**
2007	105	192	1.83
Total	**820**	**2044**	**2.49**

Note: These cases were <u>not</u> registered with Alberta Vital Statistics because they were <20 weeks gestation or weighed <500 grams at birth.